Perspectives in Genetic Counseling
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President's Beat

"You're too smart to be a genetic counselor!"

I will never forget a geneticist saying this to me in grad school. Granted, that was several years ago now, but I remember the complicated feelings her statement raised for me. I'll be honest, my initial reaction as a timid grad student was, "Thank goodness, she doesn't think I'm dumb." Then, as my thoughts continued to reel, my relief morphed to anger. Why would she, or anybody, think genetic counselors were not necessarily "smart" or not as "smart" as someone like her, a physician? What does that mean, "too smart to be a genetic counselor?" If I am smart and interested in genetics, patients, and healthcare, would I have been more useful if I'd gone to medical school? Why did my chosen career path surprise her, if I was also intelligent?

The memory of this comment resurfaced when I read an article titled, "Doing Math is for Ugly Girls" (http://thesocietypages.org/socimages/2011/05/26/doing-math-is-for-ugly-girls/). What does this title say about our society and the education of women? I didn't become a genetic counselor because I really wanted to be a doctor but couldn't cut it. I am proud of our profession and all of the highly intelligent women and men who are contributing their talents to genetic counseling.

Throughout my career I have met and interacted professionally and personally with a wide variety of medical and non-medical personnel. I admit I see intelligence as an admirable characteristic, but I value this in all individuals in all walks of life and chosen career paths. I have been stunned into silence (and believe me, that's hard to do!) by brilliant, insightful comments from "smart" ten-year-olds, devastated mothers and fathers, and the janitor one morning in the genetics building. On the other hand, uninformed, uneducated, and just plain wrong statements from physicians and other healthcare providers have also shocked me.

But I have never been *surprised* by a genetic counselor's intellect. If anything, I have felt genuine pride and amazement at the scope of my fellow colleagues' intelligence and insights into the application of our unique knowledge. If I need to call a laboratory for testing information, I automatically ask for the genetic counselor. If I am looking for a creative and different approach to solve a problem, there is always a genetic counselor whose ideas are vast, diverse, and unique. Throughout these past few months as President of the NSGC, the breadth and scope of talent I have witnessed by genetic counselors as part of Task Forces, Committees, or someone who just emails with a new idea, have been staggering and humbling.

Not so long ago, I was talking with one of the Pediatrics residents rotating through our busy general genetics clinic. She was interested in learning more about genetic counselors and what we do because she'd never heard of us (imagine that!). At the end of our conversation she told me she wished she'd become a genetic counselor. I couldn't help but smile a little as she shared her conclusion after learning about our exciting profession. Of course she did!

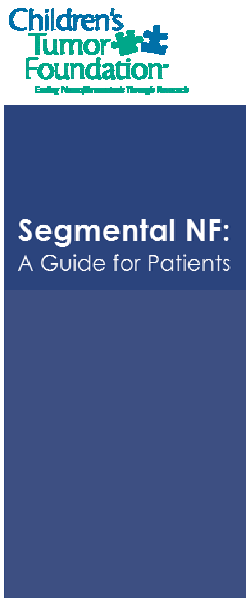
As this year draws to a close and we transition into the chaos that winter often brings (holidays, final exams, new semesters, busy clinics, etc.), start thinking about what you bring to this profession. Think about how your volunteer, educational, and academic efforts impact others' images of this profession. Consider that your interactions with colleagues, patients, and friends will impact their knowledge of and experiences with your chosen profession.

And maybe, along the way, remind yourself once in a while that you are too smart *NOT* to be a genetic counselor!

Karin M. Dent, MS, LCGC
2011 NSGC President

Overview of segmental neurofibromatosis type 1 and mosaicism: Our process and guidelines for developing a patient educational brochure

By Christina B. Hurst, MS, CGC, Alicia R. Gomes, MS, CGC and Bruce R. Korf, MD, PhD



We initiated a specialty care clinic for patients with neurofibromatosis at The University of Alabama at Birmingham in 2003. The clinic provides care for children and adults with all forms of neurofibromatosis. One of our goals is to provide our patients and families with verbal and written information. Specifically, we want to provide them with educational resources to take home and share with family, if desired. In 2009, it came to our attention that we did not have good, current resources for patients with segmental neurofibromatosis type 1 (NF1).

A review of segmental NF1 and mosaicism

In individuals with segmental NF1, the manifestations are limited to a portion of the body. This is attributed to somatic mosaicism for an *NF1* gene mutation. To explain segmental NF1, it is necessary for patients to understand the concept of mosaicism. In NF1, mosaicism can result in a milder generalized case or it can result in a segmental phenotype. It is posited that the later in development that somatic mutation occurs, the more likely the phenotype is to be segmental (Colman *et al.*, 1996; Wu *et al.*, 1997).

Several examples of both phenotypic and molecular evidence of mosaicism in NF1 have been reported; some of them describe segmental presentation of features (Listernick *et al.*, 2003; Tinschert *et al.*, 2000; Messiaen *et al.*, 2011). When mosaicism involves cells in a parent's germline, there may be an elevated risk of transmission of "full" NF1 to children. This is exemplified in the case described by Lazaro *et al.* (1994), in which mosaicism was documented in the sperm of an asymptomatic father who had two children with "full" NF. This same risk cannot be ruled out in patients who present with a segmental phenotype.

The prevalence of mosaicism in NF1 has been estimated to be <1% (Wolkenstein *et al.*, 1995). This may be underestimated due to milder symptoms in some patients that might go unrecognized, or symptoms thought not worthy of bringing to medical attention (Ruggieri and Huson, 2001). The ability to find molecular evidence of mosaicism in NF1 is affected by the type of mutation/deletion present and the testing methodology used (Messiaen *et al.*, 2011). For example, Messiaen *et al.* demonstrated that approximately 10% of patients with an *NF1* gene deletion are mosaics, typically in sporadically affected patients.

Our motivations to create a brochure

Genetic counselors often use or create visual aids to help educate their patients. Our experience with patients who demonstrated a segmental NF1 phenotype was that it was hard for them to comprehend the concept of mosaicism. We also found that there were many different clinical scenarios that would prompt the discussion of mosaicism. For example, we followed patients with plexiform neurofibromas (a type of tumor found in approximately 30-50% of NF1 patients) who did not qualify for clinical trials because the distribution of the neurofibromas was a manifestation of segmental NF1, not "full" NF1. We also saw patients who met full clinical diagnostic criteria, but only within a restricted region of the body.

We began to appreciate the psychological importance of confirming the diagnosis of segmental NF1 for patients via tissue or tumor testing, and the impact that these test results had on management of the symptoms and patient decision-making. We felt that an educational brochure that described the spectrum of findings in segmental NF1, as well as its origin, might help patients better understand their condition and its implications.

The process

In 2010, as a member of the Children's Tumor Foundation (CTF) NF Clinic Network, we were given the opportunity to apply for funding to support our efforts. One of our goals was to create a brochure for the population of segmental NF1 patients. We noted that there were not current, patient-friendly resources on segmental NF1 available through CTF or other organizations. We described our desire to create an updated brochure about segmental NF1 and to provide this to CTF for distribution. The costs of our effort were mainly limited to publication fees and professional time for the development of the brochure.

We involved our local NF clinic staff to provide their expertise, and we collaborated with staff at CTF. The aim was to answer the most common questions about segmental NF1 – including how it happens, how the diagnosis is made, the concept of mosaicism, potential complications of segmental NF1, how patients should be followed, and the risk that a person with segmental NF1 can have a child affected with "full" NF1. We chose wording and created figures that we thought would be easily understood by a general reader. In addition, we obtained permission from a patient whose clinic photographs we believed were good illustrations of the concepts. We also set a timeline to finish our project.

Guidelines we identified for developing a brochure

1. *Identify a need/rationale*

    - What materials do not exist that you could use in clinic?
    - What population is lacking an understanding of a topic, phenomenon, or condition?
    - What concepts are difficult to explain, or fundamental for patients to grasp?
    - What are common conditions in which a broad resource would be widely used?
    - What are unique populations (such as those speaking a foreign language, teenagers) that could benefit from such a resource?
    - What concept(s) are important for the general community to know about your clinic?

2. *Identify a target audience*

    - Patients
    - Referring healthcare providers
    - Family members
    - Community leaders
    - General public

3. *Identify experts or a working group*

    - Physicians
    - Genetic counselors
    - Other healthcare providers
    - Researchers
    - Laboratory directors/scientists
    - Patients
    - Professional groups or agencies
    - Advocacy groups

4. *Identify topics needed to address*

    - Description of condition
    - How to order testing
    - Goal of a specialty clinic
    - Resources available
    - Research recruitment

5. *Identify other resources already available*

    - Do similar materials already exist?
    - Is the material current?
    - Is the material comprehensive?
    - Does the material target the same audience?
    - Is the material available to a limited group?

6. *Identify graphics/photos needed*

    - Obtain and document permissions
    - Design or create figures/charts

7. *Identify format(s) to be used*

    - Website
    - Brochure
    - Event/lecture

8. *Identify timeline*

    - Variable

9. *Identify funding sources*

- Support group or national agency
- Hospital funds or donations
- Laboratory who desires an educational tool
- Research group for use in recruitment

10. *Identify publication sources*

- Local printing businesses
- Agency with advocacy resources
- Hospital printing services

11. *Identify means of distribution*

- Post to websites
- Use as a clinic handout or provide to multi-disciplinary clinics
- Provide to referring physicians/offices
- Support Groups
- Meetings (local, regional, or national)
- Other

Conclusion and challenges

Our goals for developing this brochure were 1) To create a user-friendly resource to answer common questions about segmental NF1 from a patient perspective and 2) To make this resource available to patients/families through the resources of CTF. We found CTF to be open to this idea, cooperative in the process, and able to provide funding for our effort and printing needs. Our guidelines and experience may help others with similar goals to establish a quick and task-oriented approach to getting this done.

Given the confines of any written material with a certain goal or audience in mind, it is not possible to address all topics desired. As an example, in our clinic we are frequently confronted with the difficulty of confirming the diagnosis of segmental NF1 – often from providers who are not sure how to arrange testing on non-blood specimens. While the logistics of organizing genetic testing was not a topic that we chose to discuss in our brochure in detail (as it was not aimed at healthcare providers), we did provide our contact information for those who had more questions on this topic in hopes that patients would find this helpful if needed.

How to view our brochure

To view our brochure on segmental NF1 or to access additional resources on NF, please visit The Children's Tumor Foundation website at: http://www.ctf.org/Living-with-NF/patient-information-brochures.html.

Acknowledgements

The authors are grateful to The Children's Tumor Foundation for encouraging and supporting this work.

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For Your Practice

INTRODUCING…

LEXIGENE

The French-English Genetic Counseling Lexicon

By Rachel Vanneste MSc, CGC, CCGC, Mireille Cloutier, MSc, CGC, CCGC, Julie Hathaway, MSc, CGC, CCGC and Guillaume Sillon, MSc, CCGC

What is Lexigene?

To facilitate the provision of genetic services in both of Canada's official languages, and to contribute to the development of the genetic counseling profession, a French-English lexicon of terms was created by genetic counselors for the specific purpose of aiding with genetic counseling. Trainees in bilingual genetic counseling training programs, as well as French-speaking genetic counselors who have access to the mainly English scientific literature, are in particular need of such a tool. While a few French-English medical genetics glossaries exist, none were created specifically for the field of genetic counseling and, therefore, lack many of the terms relevant to this profession.

The creation of Lexigene was funded through the Small Project Grant of the Canadian Association of Genetic Counsellors (CAGC), which was awarded to a team of four bilingual genetic counselors – **Mireille Cloutier**, **Julie Hathaway**, **Guillaume Sillon**, and **Rachel Vanneste**.

The terms in the lexicon were first gathered through six main resources where French-English translations already existed. The authors of the various resources were then contacted, in order to obtain permission to use their respective works. Terms that were not found in the original sources, but felt to be relevant by the team, were added to the master spreadsheet of terms. In total, over eighty other documents (including books, online documents from recognized websites, and specialized dictionaries) were used to populate the lexicon and/or confirm a translation. Every translation was also verified for accuracy by the four authors.

This bilingual website, www.lexigene.com, allows you to search for genetics-related terms in either French or English, and find the equivalent term in the other language. Lexigene is intended for use by genetic counselors, geneticists, and others who work in the field of genetics in both English and French on an international scale. This online tool boasts over 3,500 translated terms, with more being added regularly.

How does Lexigene work?

- Fully bilingual website (English/French) with an easy-to-use "word search" function
- Results page not only shows the "exact match" for the searched term, but all phrases that contain that word
- "Did you mean?" function provides a best guess as to what word you are trying to find when alternate spellings are possible
- Helpful hints are provided to facilitate word searching
- Browse for words in one of twelve categories
- Download a PDF that lists all the words in a specific category alphabetically in French or in English, for offline use and reference
- Feedback button creates an email form that allows you to ask a question, point out an error or suggest a term that is missing

Bookmark www.lexigene.com today or click on the Lexigene logo from the CAGC homepage at www.cagc-accg.ca

Feel free to contact the creators and administrators at admin@lexigene.com with any questions.

CAGC-ACCG

Licensure / Billing & Reimbursement

Coding Corner

Do Insurance Companies *Really* Credential Genetic Counselors?

By Bonnie Liebers, MS, CGC, Janet Williams, MS, CGC, Shanna Gustafson, MS, MPH,CGC, Monica Marvin, MS, CGC, Leslie Cohen, MS, CGC and John Richardson, NSGC Government Relations Director

Coding Corner is supported by the Coding Subcommittee of the National Society of Genetic Counselors' (NSGC) Access and Service Delivery Committee and aims to assist NSGC members with the application and understanding of governmental regulations and guidelines regarding terminology and Current Procedural Terminology (CPT) /

International Classification of Diseases (ICD) coding in genetic services as well as keep the membership educated regarding billing and reimbursement issues.

Credentialing provides a level of confirmation by an institution or a payer that a healthcare provider has met the preset qualifications to provide quality and competent care. As each institution or payer may be different in what qualifications they evaluate or what form of recognition for competencies they will assign, it is important to establish an open communication with your institution and payers. It is not uncommon for questions and uncertainties to arise when a non-licensed professional, like a genetic counselor in a state without licensure, approaches a payer or institution about credentialing. Genetic counselors might not fit neatly into a payer or institution's pre-existing categories of healthcare providers.

This can be a frustrating and confusing journey! For example, some insurers will request a genetic counselor go through an entire application process, only to inform him/her several months later that they actually do not credential genetic counselors. For this reason, do your homework before submitting an application.

- **First, find out whether the insurer or institution has an existing policy that will help them address your application.** If they don't, continue to ask questions and find the right person to help consider your request.

- **Next, explore whether or not there is a mechanism for billing that will help you to be reimbursed in the interim (e.g., out-of-plan authorization, facility fee billing, or special contract).**

- **Continue to pursue the matter of credentialing with payers (e.g., insurers) if only to educate them, even if it means re-contacting them on a regular basis to do so.** Most insurers are already paying for genetic counseling somehow, whether it is from physician billing for genetic counselors or facility fee billing. They need to be educated on the fact that they can be doing this in a less costly, more accurate, and more efficient manner. A point to be made in discussions with insurers is that without readily available genetic counseling, many physicians will go the route of more costly genetic testing to respond to patients' concerns.

Many insurers and hospitals subscribe to a national system, the Council on Affordable Healthcare Quality (CAQH). CAQH maintains credentialing information for many physicians and healthcare providers. Soon, through the work of the NSGC, genetic counselors will be able to obtain credentialing through this system, which should help when working with insurers that are CAQH members.

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

SIG Speak

From the Education Special Interest Group

Embracing the Role of Education

By Kelly East, MS, CGC, Education SIG Chair 2011

One part of what attracted me to the profession of genetic counseling was the role genetic counselors have in educating patients about the impact of genetics on health and disease. It is crucially important for our patients, many of whom have been personally impacted by genetic disease, to be given the information and support necessary to make informed decisions and become self-advocates.

There is a real need for genetics education beyond the traditional genetic counseling setting. As primary care medicine becomes more and more influenced by genetic information, everyone is going to need a basic understanding of genetics in order to be informed health consumers. In addition, numerous studies have shown that many of our colleagues in other healthcare professions feel ill equipped to integrate genetics into their everyday practices.

Genetic counselors not only have genetics expertise, we are also trained to be excellent educators. We have the knowledge and skills needed to have a large impact on the genetic literacy of our patients, the public, and other healthcare professionals.

The Education Special Interest Group (SIG) is a place for genetic counselors that work in, or have an interest in, genetics education to meet one another, share ideas, and collaborate on projects. The purpose of the Education SIG is to promote communication between genetic counselors whose work or interests include the education of patients, healthcare providers, the public, the next generation of genetic counselors, and students of all ages. In addition, the Education SIG helps promote and support the role of all genetic counselors in education. I have had the pleasure of being the Education SIG chair for 2011, which has involved helping to get the SIG up and running. I have really enjoyed getting to know other genetic counselors interested in genetics education, and I am excited to see the projects we are planning come to fruition.

I work in the Education Outreach Department at the HudsonAlpha Institute for Biotechnology in Huntsville, Alabama. Our department is responsible for providing educational content and resources for learners of all ages to better understand genetics concepts, from elementary aged students all the way up to senior citizens. We are actively involved in producing digital education resources, curriculums, and hands-on activities that are used in classrooms across the state and country. Each summer we host week-long

middle school biotechnology camps and an intensive two-week teacher academy. We also manage an institute-wide internship program, and host evening educational courses for the general public twice per year.

In addition to introducing the new Education SIG, I would like to introduce a few fellow SIG members and highlight some of the unique ways genetic counselors are getting involved in education.

Shelly Bosworth, MS, CGC, Education SIG Co-Chair

It isn't what one would expect – teaching was the tether that allowed me to continue practicing clinical genetic counseling. I graduated from the University of California - Berkeley program bright-eyed and optimistic. However, one year after I was hired by a large hospital, I got laid off.

Working as a pharmaceutical representative paid my mortgage, but was dull. I wanted to get back on my intended course. Surveying the opportunities in Eugene, Oregon I decided to write a letter to the community college. I wrote: "Lane Community College should have a genetics professional on staff." I told them why my training as a genetic counselor qualifies me to teach genetics to undergraduates, and why having me on staff would benefit the college. They called! I taught Genetics, followed by Introductory Biology, Introductory Physiology, and a few other classes. The pleasure of introducing genetics to hundreds of minds is immense. Even better, while teaching was keeping me connected to science, my previous employer offered me a part-time genetic counseling position. I was back on course!

Now I teach four Genetics classes per year at Lane Community College, and work half time as a prenatal genetic counselor. I have job security, and I love both roles. It is a wonderful combination because each role benefits the other. My students ask questions to keep me researching and learning topics that go beyond prenatal genetics dilemmas, which have become a bit routine. If I did not teach, I think prenatal genetics would start to feel like a rut. Instead, I hope and expect to keep up both roles for years to come. I love splitting my time between prenatal patients and students.

Ana Morales, MS, CGC

My interest in education is continually fulfilled in my daily work. I became involved in genetics education when I joined the University of Miami (UM) in 2006, following a request from Puerto Rico's Ponce School of Medicine for medical genetics instructors to teach their first year class. I was approached to join other UM faculty in the project, and a telemedicine course was created from scratch.

The experience demonstrated that genetic counselors' abilities to demystify and share complex information provides an advantage for teaching genetics. The experience ignited

my interest in education and launched a string of opportunities, such as teaching locally at UM School of Medicine, spearheading expanded newborn screening in-services in Puerto Rico and the Virgin Islands, and most recently, joining the Education SIG.

Public interest in our field has exploded. As a developing field, medical genetics is in need of answers to innumerable questions, but there aren't many genetics professionals to help solve them. With more genetic counselors in an educator role, we can help shed some light on these issues. I believe reaching that critical mass begins with education.

Nancy Steinberg Warren, MS, CGC

I attended college at State University of New York Stony Brook as a tried and true member of the "Boomer" generation, and graduated as a Biology major with certification in secondary science teaching. I worked for one (dismal) year as a seventh grade science teacher in a suburban Long Island community. In my student teaching experience, I learned how to teach science to motivated students, not how to discipline disinterested kids. I faced the tough realization that school teaching was not a good fit for me.

Thankfully, I learned about the new field of genetic counseling from one of my professors, Dr. Bentley Glass, and I began the Human Genetics Program at Sarah Lawrence College the following year. I liked, and was much better at, teaching people who really wanted to know more about their family history, birth defects, genetic conditions, and options for testing and support. In every professional position I held, I gravitated toward new educational challenges, including providing education to healthcare professionals, students, and the public, and developing curriculum materials, activities, and resources.

Most recently, I was awarded the 2009 Jane Engelberg Memorial Fellowship (JEMF) to learn more about the role of cultural diversity and cultural competence in reducing health disparities, and also to hone my knowledge and skills in pedagogy. The outcome of this JEMF project is the Genetic Counseling Cultural Competence Toolkit (http://www.geneticcounselingtoolkit.com), a website where I apply what I learned to genetic counseling cases, which are approved for self-study and Category 1 Continuing Education Units for genetic counselors. The site is also a repository of resources and links to help our profession take advantage of the rich experience of other fields.

My current educational efforts focus on preparing and delivering on-site workshops on "building cultural capacity" to genetic counseling training program staff and students, genetics service provider organizations, and genetics companies. My identity as an educator is synonymous with my identity as a genetic counselor.

Do you have an interest in education? Join us – we would love to have you as a SIG member! Contact us at keast@hudsonalpha.org or shbosworth@womenscare.com.

NSGC News

By the NSGC Executive Office

National Society of Genetic Counselors (NSGC) Committee Updates

Have you wondered what type of activity is taking place within the NSGC's five management committees? Below, you will find an update on many exciting activities that were underway during the second and third quarters of 2011. Updates will be presented in *Perspectives in Genetic Counseling* twice annually to keep you informed and help you determine where you might want to get involved!

Access and Service Delivery Committee

Monica Marvin, Chair
Shanna Gustafson, Vice Chair

- In collaboration with the Public Policy Committee, the Payer Task Force presented the webinar, "Preaching to the Choir: How to Prove the Value of Genetic Counseling in a Competitive and Evolving Healthcare Landscape" on September 28, 2011. The recording of the webinar is available on the NSGC website http://www.nsgc.org/Education/Webinars/tabid/440/Default.aspx.
- The Payer Task Force is developing a document titled, "Elements of Genetic Counseling," to be used in discussions with payers.
- The Licensure Subcommittee continues to provide support for licensure efforts in individual states.
- The Service Delivery Models Task Force continues to work on analysis of a 2010 survey of the NSGC membership to identify existing and innovative models for delivery of genetic counseling services.
- The Practice Guidelines Subcommittee has been converted to a stand-alone Committee as of September 2011.
    - The Practice Guidelines Committee continues to foster the development of multiple practice guidelines. The Committee has one practice guideline in press titled: "NSGC Genetic Cancer Risk Assessment Guidelines: Recommendations of the National Society of Genetic Counselors," which will be published in the *Journal of Genetic Counseling*.
    - The NSGC Board of Directors has approved eight additional guidelines, which are in various stages of review by the Practice Guideline Committee.

    - The Committee is working to partner with the Special Interest Group (SIG) leadership to encourage more practice guidelines, especially in the areas of referral indications for genetic counseling.

- The Committee developed an online Credentialing Course, approved for 1.0 CEUs, which launched in July 2011.
- Regular submissions are made to *Perspectives in Genetic Counseling* for "Coding Corner." Frequently asked questions about coding are highlighted for the benefit of the membership.

Communications Committee

Amy Sturm, Chair
Kimberly Barr, Vice Chair

- Finalized a one-page document to be used by the NSGC's leadership and staff to ensure consistent communication of the NSGC's brand in all external communications.
- Working with SIGs on development of specialty-specific website content, including developing information for healthcare providers. This information will be available in late 2011.
- Implemented new NSGC Discussion Forums, moving from the listservs to this new communication platform offering enhanced technology.
- An assessment of current publications available from the NSGC is underway. Recommendations will be made regarding updates/edits/removal, or new publications that are needed.
- Launched the first phase of the NSGC self-marketing tool kits on the NSGC website. The tool kits are a customizable resource available for the NSGC members to use in marketing themselves to referring healthcare providers. Additional information, such as specialty-specific content, will be added to the toolkits on an ongoing basis.
- Incorporating a new column in *Perspectives in Genetic Counseling* about the latest trends in health industry and technology. This column will begin in 2012.

Education Committee

Julianne O'Daniel, Chair
Leigha Senter, Vice Chair
Renee Chard, Interim Chair

- The Annual Education Conference (AEC) was held in San Diego, California October 27-30, 2011.

- The Webinar Subcommittee held six webinars in 2011. The most recent webinar was held at noon C.S.T. on November 30, 2011. It reviewed the latest research in Fragile X carrier testing.
- The Webinar Subcommittee is working on webinar topics for early 2012.
- The 2011 online course, "Genomics and Personalized Medicine," was launched in July 2011.
- The Outreach Education subcommittee has worked with the Cancer SIG to develop a proposal for outreach education with the Association of Community Cancer Centers (ACCC), a multidisciplinary association targeting all members of the cancer care team. The Cancer SIG will work with ACCC to implement this proposal on behalf of the SIG.
- The Outreach Education Subcommittee is soliciting new proposals from the SIGs for development of an outreach education program with a physician or referring healthcare provider organization in 2012.
- To keep up with the demand for reviewing both Category 1 and Category 2 Continuing Education Unit (CEU) applications, the CEU Subcommittee added three review teams in 2011, for a total of 15 review teams.
- The *Journal of Genetic Counseling* CEU program is well underway, with a final registration deadline in November 2011.

Membership Committee

Samantha Baxter, Chair
Bronson Riley, Vice Chair

- Conducted a very successful "First-Time Attendee and Student Orientation" at the 2011 AEC in San Diego, in conjunction with the Student/New Member SIG.
- The Organizational Cultural Competency Task Force collaborated on the Diversity Preconference Session at the 2011 AEC.
- The Awards Subcommittee administered the 2011 Leadership Awards program; all awardees were honored at the 2011AEC.
- The Awards Subcommittee administered the 2011 Cultural Competency Scholarship program. Two students received scholarships to support their AEC attendance.
- Launched a new cycle of the NSGC Mentorship Program in June 2011, and prepared for the November 2011 session launch.
- The Leadership Task Force held a pilot Leadership Development Session at the 2011 AEC for all incoming NSGC leaders. A follow-up teleclass will be offered to all session participants in early 2012.
- The Professional Status Survey (PSS) Subcommittee has started work on the 2012 PSS. Survey revisions are near-final, and an online survey administrator has been selected.

Public Policy Committee

Susan Hahn, Chair
Flavia Facio, Vice Chair

- Presented "The Evolving Landscape of Genetic Testing Oversight: Gaps from the Past, Present Status, and Potential Implications for the Future" at the 2011 AEC.
- Presented the webinar, "Preaching to the Choir: How to Prove the Value of Genetic Counseling in a Competitive and Evolving Healthcare Landscape" on September 28, 2011, in collaboration with the Payer Task Force of the Access and Service Delivery Committee. The recording of the webinar is available on the NSGC websitehttp://www.nsgc.org/Education/Webinars/tabid/440/Default.aspx.
- Completed the NSGC Position Statement on Healthcare Reform
- Completed the revised NSGC Position Statement on Nondiscrimination
- Completed the revised NSGC Position Statement on Direct-To-Consumer Genetic Testing
- Position statements in development include:
    - Newborn screening
    - Blood spot storage and use
    - Non-invasive prenatal diagnosis
    - Stem cell research
- Reviewing and updating the NSGC's bibliography for distribution to legislators in support of the NSGC's proposed federal legislation
- Letter to the Editor was submitted and accepted *Journal of the American Medical Association* (Buchanan AH and Stopfer JE. Genetic counseling in oncology. *JAMA*. 306(13):1442; author reply 1442-3. 2011.) highlighting the fact that genetic counselors should be recognized providers under the Centers for Medicare and Medicaid Services (CMS), in response to the 2011 article by Tung N (Tung N. Management of women with BRCA mutations: a 41-year-old with a BRCA mutation and a recent history of breast cancer. *JAMA*. 305(21):2211-20. Epub 2011 May 10).
- Assessed the NSGC's 2011 liaisons and made recommendations to the Board for liaison relationships to continue or pursue in 2012.

ABGC Update

Activities of the Accreditation Committee

By Janice Berliner, Outgoing Chair and Cecelia Bellcross, Incoming Chair

The Accreditation Committee of the ABGC has been very busy this year, responding to the needs of our diplomates and programs and supporting our profession. In early 2011 we initiated a Task Force to complete a comprehensive review and revision of the document entitled: **Required Criteria for Graduate Programs Seeking Accreditation by the American Board of Genetic Counseling.** This document specifies the minimum standards used by the ABGC to accredit Genetic Counseling Training Programs.

The original document was adopted in 1996, and has undergone periodic updates, but no comprehensive revisions. The Task Force completed Phase I of its charge, which was to perform a detailed review, compare the document with published standards of other comparable professions (e.g., Physician Assistants), and propose revisions for board approval.

The Task Force is currently in Phase II of the process: completing the revisions for early 2012. The revisions include major reorganization, streamlining the document, eliminating redundancy, content-specific changes to reflect the current practice of genetic counseling based on the recent Practice Analysis, and updating of the Practice-Based Competencies (see below). In addition, the "Required Criteria" are being renamed "Standards" to be consistent with those of related professions. The ABGC wishes to acknowledge the hard work and dedication of the Task Force members: **Nancy Steinberg Warren** (Chair), **Cecelia Bellcross** (Board Liaison), **Sharon Aufox**, **MaryAnn Campion**, **Vera Cherepakho**, **Sarah Noblin**, **Kate Reed**, and **Colleen Schmitt**.

We also reviewed the Practice Based Competencies (PBCs), which are used by the training programs to design their curricula to meet minimum training standards. These were originally written in 1996 and last revised in 2006. A Task Force meeting was convened in October, and members thought strategically about what components are unique to the field of genetic counseling. They reviewed, evaluated, and began to revise the document. The Task Force members were chosen based on geography, years in practice, and specialty area, to represent our profession in a diverse way. A draft of the revised PBCs will be circulated for comments from our diplomates before being finalized. The Task Force was led by Board Liaison **Cathi Rubin Franklin** and facilitator/ABGC President **Deb Lochner Doyle**, and included members **Jehannine Austin**, **Rawan Awwad**, **Bonnie Baty**, **Amanda Bergner**, **Stephanie Brewster**, **Lori Erby**, **Anne Greb**, **Robin Grubs**, **Gillian Hooker**, **Kelly Ormond**, **Christina Palmer**, **Liz Petty**, **Claire Singletary**, **Matthew Thomas**, **Helga Toriello**, **Wendy Uhlmann**, and **Carol Walton**. The timeline for completion of the revised document is Spring 2012. Thank you all for a job well done.

A major focus of the Accreditation Committee this year has been our separation from the credentialing arm of the ABGC. It is important for an accrediting body to be separate from the credentialing body to avoid the perception of "teaching to the test." This is a well-established practice, and a goal to which the Board has aspired for some time.

The new name of the accrediting body will be the **Accreditation Council of Genetic Counseling (ACGC)**. Its mission will be to protect the interest of students and the public by setting standards for genetic counseling education and accrediting graduate programs. Further, the purpose of the ACGC will be to provide leadership by protecting the interests of the students, public and the integrity of the genetic counseling profession by:

- Establishing standards for graduate level genetic counseling education
- Evaluating educational programs to ensure compliance with those standards
- Accrediting genetic counseling training programs that meet the ACGC standards

The ACGC will have a ten-member Board and initially may be an affiliate of the ABGC before ultimately becoming a stand-alone entity. If allowed, this transitional structure will allow for a quicker transition to becoming a separate board and permit us to use our existing management contract with Applied Measurement Professionals, Inc. (AMP) through 2015. It reduces our financial and administrative risks, and provides for a quicker completion of existing essential projects. The transition Task Force has done a tremendous job, led by **Holly Peay** and including members **Lisa Amacker North**, **Jennifer Fitzpatrick**, **Kathy Valverde**, **Meredith Weaver**, and **Robin Bennett** (Board Liaison).

As you can see, the Accreditation Committee, as well as the Board of Directors in general, have had a productive 2011! Please do not hesitate to contact any Board member if you have any questions about Board activities, or to place your name on the ABGC volunteer list. For more information or contact information for Board members, please see the website at www.abgc.net.

Student Forum

What a Difference a Year Can Make

By Melanie Hardy, MS; University of North Carolina at Greensboro, Class of 2011

When I first considered writing an opinion piece for *Perspectives in Genetic Counseling*, it was the Fall of my second year in the genetic counseling program at the University of North Carolina at Greensboro. I had recently returned from the National Society of Genetic Counselors (NSGC) Annual Education Conference (AEC) in Dallas, where I had a myriad of experiences. I was so impressed by the scale of the conference, and the opportunities that were available from which to learn and become inspired. I watched as genetic counselors at all stages in their careers gave talks, presented posters, chatted in the hallways, shared information, and had reunions. I gained a new appreciation for our profession. Although genetic counselors are a rare breed in comparison to many other professions, we do know how to pack a conference center!

As I took time to reflect on my experiences at the AEC, I came to the preliminary conclusion that genetic counseling was a profession that a student like me would have to learn to crack. I felt like an outsider, because I had the impression that everyone seemed to know everyone else. This was an intimidating conclusion. All of these people were connected by years of interactions; I wanted to be part of that as well! I decided to figure out how to carve a place for myself in this tight-knit community.

As a second year student, I had plenty of other things on my mind; still, I began to seek opportunities to get involved. I heard that little nagging voice in the back of my head saying, "Are you kidding? What makes you think you have time to do more than what you're currently doing? Classes, clinic, research, graduate assistantship, job search… there's no way you have time for anything more!" The voice in my head was making a convincing argument to maintain the status quo. And I didn't listen.

Although it was a tough decision to ignore that voice, I am so glad I did! It turns out that finding opportunities to become involved is the key to becoming an insider. Going the extra mile not only builds a resume, it builds relationships. By investing a little of myself here and there, I began forming connections with genetic counselors across the country.

In case you find yourself in the same position I was in as a student, let me give you my personal recipe for success:

Join groups. I happened to choose the Student/New Member Special Interest Group (SIG) within the NSGC. At the 2010 AEC, I went to the SIG meeting with only the intention to find out what the group was all about. I ended up leaving the meeting with an application to serve as leader on one of the Task Forces within the SIG. Long story short, I am now the Co-Leader of the Outreach Task Force. Through the Task Force, I have met many wonderful people that are dedicated to the same vision: giving presentations to audiences around our area about genetic counseling as a career and a profession that is dedicated to serving patients.

As Co-Leader, I work closely with our members, SIG leaders, and other SIGs within the NSGC to plan and participate in events for the AEC. So taking advantage of just one

opportunity to attend that first meeting has opened up more new opportunities than I could have ever dreamed. It was the best first step I could have made.

Take advantage of mentor/mentee opportunities. The NSGC has regular opportunities to pair students and new genetic counselors with those that have experience in the field. I have had two mentors through this program and have gained much wisdom from our conversations.

I also have a mentor through the Student/New Member SIG who is a genetic counselor closer to my career stage than other mentors I have had in the past. Since she is newer to the genetic counseling profession, she has a great deal of knowledge about the job market, trends in hiring, and what it is like to be in the exact position I am now. Her encouragement has helped me through the ups and downs I have experienced as a second year student and new graduate. Mentors are people with a desire to help; they offer encouragement and advice. They are an incredible resource and another chance for a student to make a connection. I highly recommend taking advantage of opportunities to become a mentee.

Seek out hidden opportunities. These are the "hidden opportunities" disguised as extra work. For example, at one of my clinical rotations, the doctors and counselors were working on developing a new disorder-specific clinic to be opened soon after I finished my rotation. Although there was much work to do, one thing that had stalled was the development of disorder-specific, patient-friendly materials. Although it was not a requirement for my rotation, I decided to work on the development of these resources.

By the end of my rotation, I was able to work closely with the counselor who was developing the clinic and received feedback from the supervising physician. If I had not done the work, I would have had only a minimal chance to work with either of these professionals and would have missed an opportunity to get some insight into the work involved in opening a specialty clinic. Working relationships such as these have the potential to open doors in a myriad of ways. They allow a student to stand out as more than just someone who completed a rotation. Not only did I learn from the work I did, I have also formed more connections in the field.

Put yourself out there. I was fortunate to have Program Directors that diligently sent us information on any opportunities that came along. Whether it was hanging out with kids while their parents were in a meeting about 22q deletion syndrome, volunteering at the local Down Syndrome Society fundraising event, or talking about genetics at a local elementary school, I tried to get involved. Through these activities, I met many of the providers that work locally and are well connected in the genetics community. Although each provider may not remember me from one activity or one day of volunteering, you never know where those opportunities might lead.

I even worked as a DNA Day expert by participating in online question-and-response through the National Human Genetics Research Institute. Talk about putting yourself out there! I wasn't sure I had anything to offer to students and teachers who posted questions,

but I am glad I did it. Being involved gives me the sense that I am an active member in a community of genetics professionals.

As I write this article, I am approaching the challenge of fitting into the genetic counseling profession from a completely different perspective. I have learned that not all genetic counselors know each other. They have all gone through the same struggles I have, and they found their way into the profession by getting involved. Not everyone did so in the same way, but they each found their niche. There are plenty of opportunities to fit in; it just takes a little effort and a willingness to share yourself and your talents with others.

The New Graduate Life

The Morphology of My Canadian Accent

By Cherise M. Klotz, MS, CGC, CCGC

I'd always assumed I would return to my home province in Canada to work as a genetic counselor after completing my graduate studies in Pittsburgh. When graduation finally approached, however, the right job was not available in the right place – and I quickly realized I might have to move forward with an alternate plan. Plan B was to job-search in other large Canadian cities and to consider a selection of cities in the U.S. After a number of job offers, some heartache, a lot of hope, countless conversations with supportive friends, and finally a work visa, I accepted a genetic counseling position in a mid-sized city in West Michigan.

If someone would have told me years ago that my first job as a genetic counselor would be located in West Michigan, I would have guffawed in disbelief. Although I grew up in the Canadian prairies, I'm a big-city girl. I left small-town life at the age of seventeen and never looked back.

I knew no one in Michigan. I love the arts; I needed culture. I love the mountains; I needed altitude. I needed family. It took me some time to realize that, hidden at the core

of all these “needs,” perhaps I was simply looking for a challenge. And I certainly acquired a challenge.

I learn by being thrown into a situation. When I started my job in Michigan, I was the only genetic counselor in a high-volume Maternal-Fetal Medicine clinic with four perinatologists. Very quickly, I gained professional autonomy. I learned an incredible amount about cultural competency and working with translators. I collaborated with other genetics professionals and lab genetic counselors in a way I may not have experienced in Canada. I was close enough to a number of tertiary care centers that some of my patients were participating in groundbreaking clinical studies. It felt as though I was in the middle of all the excitement.

I worked in Michigan for a year-and-a-half until my personal life brought me back to my Canadian roots. I remember my Canada-to-U.S. transition as being much more difficult than I expected it to be. But in the end, I said goodbye to a job and a city that I’d learned to love. I also had an apartment full of furniture I didn’t want to sell (and therefore had to move) and had to import my American-bought vehicle into Canada. I had to re-learn what it meant to work in a unionized environment, and had to adapt to having my family close by again. But on a positive note, the major advantage of my move back to Canada was an improvement in my work/life balance. My life in Michigan was heavily focused on my job; moving back home to Alberta made it easier for me to focus on other aspects of my life as well.

Friends, family members, and colleagues have often asked me which healthcare system I like better – the American or the Canadian. I have always answered this question the same – there are benefits and drawbacks to both systems. Of course, being a genetic counselor, I tend to weigh the pros and cons.

In Canada: a socialized healthcare system that allows all residents access to genetic counseling services and oftentimes genetic testing, provided that the testing is medically indicated. Justice, equality, opportunity. At the same time, however, in Canada some genetic tests can take a year or more before the clinician has the results in hand, and some clinics have year-long (or more) waiting lists due to limited resources.

In the U.S.: those with good health insurance are often very well taken care of, particularly those who have insurance plans offered at reasonable costs through their employer. Genetic testing is relatively fast and wait lists are often negligible. Timing can be everything. Still, for example, I saw a number of patients who were limited in the healthcare they could access during pregnancy because of their limited insurance coverage.

I remember living in America and wondering, “How might this patient’s situation be different if she were living in Canada?” Now that I have returned to Canada, I have occasionally wondered, “How might this patient’s situation be different if she were living in America?” After weighing this back and forth in my head countless times, I have concluded that neither system is the “right way.”

Another interesting challenge I faced as a Canadian genetic counselor working in the U.S. was regarding professional certification. Throughout graduate school, I was a member of both the National Society of Genetic Counselors and the Canadian Association of Genetic Counsellors (CAGC). I maintained both those memberships as I began working as a new genetic counselor. Because I was working in America and I graduated from a U.S. genetic counseling program, I knew that I wanted to have certification by the American Board of Genetic Counseling (ABGC).

I am a Spring 2009 graduate and my classmates were taking the ABGC exam in Summer 2010, so I joined them. Although the ABGC exam is now offered every year, the CAGC exam is still offered every other year; it was unfortunately not being offered in 2010, when I sat for my ABGC board exam. I know some genetic counselors that were in similar situations as I, and they waited until 2011 – to take both exams in the same year.

Admittedly, I felt pressured to take my ABGC exam in 2010. So, for better or for worse, I was faced with the challenge of studying for the exams one year apart. Happily, it concluded well. I was able to celebrate two years in a row, after passing my ABGC exam in 2010 and my CAGC exam subsequently in 2011.

I know a number of Canadian genetic counselors that have attended graduate school in the U.S. and have returned home to Canada immediately after graduation to work. I also know several that have worked in the U.S. and stayed there long-term. I, however, worked in the U.S. for a short while, and then came back to Canada. This was the best option for me and I believe that it helped me grow both personally and professionally.

When I lived in Pittsburgh for graduate school, locals used to (ironically) ask me if my accent was from Michigan. When I lived in Michigan, many guessed that I hailed from "The Great White North." Now that I am back in the Albertan prairies, I catch myself saying phrases with an American accent every once in a while. I've considered all the cities I have lived in to be "home" at some point. And I'm sure that my accent will remain somewhere in between.

Genetic Counselor Publications

Feature Article

By Christine Colón, MS

Reis LM, Semina EV. Genetics of anterior segment dysgenesis disorders. *Curr Opin Ophthalmol.* 2011;22(5):314-24.

Linda Reis, MS, CGC

There was a time in the not-so-distant past when being a genetic counselor in a research-based position was classified as having a “non-traditional” role. Typically, after graduating from a training program, many would take clinical genetic counseling positions. However, as our world continues to change, so does our need for knowledge and its potential applications within the field of genetics. Today, more genetic counselors are working in research settings and are specializing in highly specific research areas. One such counselor is **Linda Reis, MS, CGC**, who is part of a team specializing in discovering the genetic causes of both isolated and syndromic eye disorders.

Linda was always interested in research, and has been actively participating in studies since her completion of The University of Cincinnati Genetic Counseling Program in 2004. After graduation, her search for a job in the Midwest led to the offer of a dual clinical and research position at the Children’s Hospital of Wisconsin and Medical College of Wisconsin (as study coordinator for Dr. Elena Semina). She spent the next four years in these roles until she elected to work part-time, focusing on the research aspect of her job. While Linda enjoys the investigative nature of her work, she also values the occasional face-to-face patient interactions she experiences when consenting local participants for the studies, or explaining results to them.

Over the last seven years, Linda and her colleagues have co-authored several papers on the genetics of eye disorders. Their most recent publication is a review of both well-documented genes and novel findings associated with anterior segment dysgenesis (ASD). Since the initial linking of *PAX6* mutations to ASD nearly twenty years ago, several other genes have also been implicated, such as *BMP4*, *CYP1B1*, *FOXC1*, *FOXC2*, *FOXE3*, *JAG1*, *LAMB2*, *PITX2*, and *PITX3*. Newly added to this list are mutations in *COL4A1* and *B3GALTL*. Recent studies have also discovered disease-causing deletions outside the coding region of *PAX6* (downstream) and *PITX2* (upstream). Based on these results, the authors suggest investigating the upstream/downstream regions of other genes for possible deletions. In addition, cases with specific chromosomal abnormalities have been associated with ASD.

ASD conditions display multiple modes of inheritance and can have great phenotypic variability, even within families. Historically, diseases of this nature present many research and clinical challenges, and limited information about prognosis and recurrence risk is available. Studies exploring ASD conditions are looking to bridge the knowledge gap and provide more useful information to patients and their families.

The studies conducted by Linda and her team at the Children's Research Institute at the Medical College of Wisconsin include participants from several different countries. “We have been able to identify a genetic etiology for many cases, thus providing a precise diagnosis and in some cases guiding clinical management,” Linda notes. Physicians from anywhere in the world can find these studies online through the GeneTests/National Center for Biotechnology Information (NCBI) and contact Linda or her colleagues to find more information. Because many diseases associated with ASD are rare, evaluating as many cases as possible helps to strengthen and solidify previous findings, and expand upon the existing body of knowledge.

Linda enjoys the highly specialized nature of eye disorders. She feels this research is important so families are able to have an explanation for what is causing their child’s disorder and related complications. Since many different genes can cause overlapping symptoms in patients, knowing which gene is responsible can help refine recurrence risk and indicate to doctors which symptoms should be watched for, and which may be excluded from monitoring. To gauge the effectiveness of the research, study participants are given the opportunity to fill out feedback forms to express their feelings and concerns about the research, which (to date) have been collectively positive. “For families, just having more information is really empowering,” Linda states. “They’re just glad that somebody is studying these rare conditions that affect their children.”

As with many genetic diseases, Linda feels the identification of causes of ASD will be furthered by advancing technology. Whole-genome and whole-exome sequencing may now be used alongside array comparative genomic hybridization (aCGH) to help reveal new answers to previously unresolved questions. These techniques bring exciting new possibilities for gene identification; still, Linda is realistic about how much information new discoveries will contribute. “Some ASDs…probably are going to be a mix, where some [causes] are genetic and some are environmental... but [in] the syndromic or

familial conditions, hopefully we should be able to use whole-exome sequencing to identify what genes are involved."

Linda admits she had limited experience with research before starting her position. She learned the specifics on the job while utilizing the critical thinking skills she honed as a graduate student. Linda offers this advice to students and genetic counselors thinking about making research part of their career: "Find a center where you can do both [clinical and research]… and approach the PhDs who are working in genetics. A lot of researchers… are interested in learning what a genetic counselor could add to their study, and are very interested in collaborating."

Articles co-authored by genetic counselors from January 2011 – September 2011

(Names of genetic counselors appear in bold)

By Jamie C. Fong, MS, CGC

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Please send references of published articles by genetic counselors to Jamie Fong at jfong@memory.ucsf.edu

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AEC Update

Save the Date for the 2012 Annual Education Conference in Boston, Massachusetts

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

As evidenced by the overwhelming number of attendees at the 30th Annual Education Conference (AEC) in San Diego, the NSGC has significantly outgrown its traditional conference format. We are thrilled to be moving to a conference center for the 31st AEC. This new venue will allow for increased flexibility in formatting and utilization of space. Please mark your calendars and plan to join us at the Hynes Convention Center in downtown Boston on **October 24-27, 2012**.

The Hynes Convention Center is located right in the heart of Boston's historic Back Bay neighborhood, surrounded by world-class shopping, dining, and entertainment. Visit http://advantageboston.com/Hynes/Advantages.aspx or http://www.cityofboston.gov/visitors/thingstodo.asp for more information on the Convention Center and things to do in the Boston area.

Two nearby hotels, the Boston Marriott Copley Place and the Sheraton Boston Hotel, will have room blocks available for NSGC AEC attendees.

AEC Timeline Shifts in 2012

In response to attendee feedback, the 31st AEC will shift from a Thursday-Sunday format and move to a Wednesday-Saturday format. With this change, we are hopeful that attendees will be granted sufficient time away from work in order to earn the important CEUs provided by the AEC, yet still able to return home in a timely manner in order to be with family and friends prior to starting their workweeks. The Pre-Conference Symposia will take place on Wednesday, October 24, 2012 and the AEC will again begin in the afternoon with the "Welcome to the AEC" orientation, followed by the opening Plenary Janus Series and Best Abstract Awards. Concluding this kickoff will be the Welcome Reception in the Exhibitor Suite on Wednesday evening. There will be two full days of outstanding educational opportunities within the Plenary, Educational Breakout Sessions (EBS) and concurrent papers on Thursday and Friday, followed by a shorter day on Saturday with the conference concluding in the late afternoon.

Call for Speakers – November 28, 2011 to January 13, 2012

The NSGC is actively inviting members to submit presentation proposals for Plenary Sessions including the **Janus series**, **Educational Breakout Sessions** and **Pre-Conference Symposia**. We are seeking informative, cutting-edge, and stimulating presentations by genetic counselors, physicians, researchers, and other industry leaders that will help advance our knowledge within the profession of genetic counseling. **The Call for Speakers is open and the deadline for submission is January 13, 2012.** Applicants will be notified of their acceptance in late February. Watch for additional information in upcoming NSGC e-mails and on the NSGC's Web site.

Submission Guidelines

AEC presentation submissions need to contain a brief descriptive paragraph outlining the proposed content (<300 words), as well as three learning objectives written to the continuing education standards. Submissions that present a well-thought-out content outline and speaker plan generally score more favorably than those that contain minimal information. Please be reminded that a Plenary Session is typically one hour, an EBS will run 90 minutes to two hours, and a Pre-Conference Symposium is typically five hours in length. The Janus series features three expert genetic counselors, each providing a 30-minute overview on their areas of expertise. Janus series submissions should be submitted under the Plenary category in the online submission process. 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 2012 AEC Planning Subcommittee will carefully review all submissions. Proposals may be considered for other session formats in addition to the format requested.

Please note: If you are submitting/coordinating the proposal but do not plan to speak, you will have the opportunity to list yourself as such. You will be considered the main contact for the proposal. You will also need to provide contact information for all of the speakers at the time of submission. *It is important that the Subcommittee knows ALL speakers for each proposal during their review, as this will impact the sessions selected.*

Submit your proposal by completing the online submission. Instructions and the submission form can be found at the following link: http://www.nsgc.org/Education/2012AnnualEducationConference/CallforSpeakers/tabid/449/Default.aspx. Questions may be directed to the AEC Chair, **Claire N. Singletary**, at claire.n.singletary@uth.tmc.edu, to the Vice-Chair, **Quinn Stein**, at quinn.stein@sandordhealth.org, or to the **NSGC** at nsgc@nsgc.org.

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

The 2012 AEC Subcommittee

Please feel free to contact Subcommittee members with ideas, comments, and suggestions:

AEC Chair

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Resources / Book Review

Reviewed by Claire Noll, MS, CGC

Genetic Rounds:
A Doctor's Encounters in the Field that Revolutionized Medicine

Author: Robert Marion, M.D.
Publisher: Kaplan Publishing (October 5, 2009)
Pages: 278
Retail Price: $24.95
ISBN: 978-1-60714-460-1

For more than thirty years, Dr. Robert Marion has been a clinical geneticist and professor of medical genetics at several hospitals in the New York City area. In the book's introduction, Dr. Marion describes his early fascination with genetics as an undergraduate, followed by "spending all my free time hanging around" with the genetic counseling staff as a medical student. He also sets the stage for stories of life as a geneticist by writing about an amniocentesis he observed during his early residency. The patient was the wife of one of his professors; the fetus had Down syndrome. This pregnancy was interrupted, and the subsequent one resulted in a baby with typical chromosomes. Much later, Dr. Marion met the couple at a banquet, at which their son and his daughter both received awards. None of them mentioned the couple's first pregnancy, or the twenty-year gap between intimate life events that were unexpectedly shared.
The sixteen stories that Dr. Marion recounts frequently mention such serendipity, but also missed opportunities for making personal connections. Serendipity allowed him to make a diagnosis for a child who had long puzzled doctors, simply because he had recently researched that disease. It enabled the parents of a previous child with spinal muscular atrophy to have the option of prenatal diagnosis by using a keepsake hair sample these grieving parents hadn't thought to mention at first; it ultimately avoided exhumation to obtain a DNA sample (prior to the availability of direct DNA testing for the condition). He is careful to explain that such fortunate luck happened within the context of good clinical care, attention to detail, listening to the parents, and staying current with technological advancements.

Dr. Marion also reveals that constraint kept him from expressing sorrow to the family of a neonate who died during a NICU shift during his residency because he was anxious to leave, and then again from addressing an astonishing and unanticipated disclosure by the father of a child who had a terminal disease at a small-group exercise for medical

students. Dr. Marion explains how exploring his reticence actually led to greater appreciation of family dynamics, and the support he could offer families by sharing his feelings.

A welcome thread running through these vignettes is the respect he shows for patients and families. Dr. Marion describes his struggle over telling a college friend at a reunion that he believed his friend's child had a genetic disorder. This expanded to frustration over recognizing that a local television personality had Crouzon syndrome, and not knowing whether he should intervene.

Additional psychosocial issues, such as understanding that there are two "patients" in pediatric genetics (the patient and the family), treating emotions as well as medical symptoms, not passing judgment when secrets are revealed, caring for a family experiencing fetal demise or childhood death well past the actual passing, and deciding whether or not to disclose abnormal results on the brink of a major holiday, will be familiar to genetic counselors. But by using his own emotions and reflections as a framework to surround challenging case studies, Dr. Marion adds warmth and complexity to what might otherwise seem like "war stories from the genetics trenches" to less-informed readers.

Indeed, this book is written for lay readers. Inheritance patterns, genetic testing, and some disease states are adequately explained for that audience. Only two things could be confusing. First, Dr. Marion makes some sweeping statements in the Introduction about the value of genetics in treating complex diseases, yet his stories describe single-gene or chromosomal disorders. Later, in a chapter concerning Marfan syndrome, he writes about aortic aneurysms and mentions Flo Hyman, Jonathan Larson, and John Ritter. While Flo Hyman had some physical manifestations of Marfan syndrome, John Ritter is known to have had a familial thoracic aortic aneurysm syndrome, and Jonathan Larson was thought to have that same diagnosis. The casual reader might mistakenly conclude that all three had been diagnosed with Marfan syndrome.

Dr. Marion's book effectively models how to write about difficult circumstances with compassion, empathy, and professionalism. His appreciation of genetic counselors is evident. This is the perfect book to offer to colleagues and patients who remark, "I don't know how you can do what you do," because it shows both the struggles and the successes encountered in clinical genetics.

Research Network

By Emily Place, MS, CGC

Early Brain Development in Disorders of the Sex Chromosomes and Disorders of Sexual Differentiation

The purpose of this research study is to learn about brain development in children with Turner syndrome (TS), beginning at two weeks of age. Participating children will have a magnetic resonance imaging (MRI) scan of the brain in the first month of life at the University of North Carolina at Chapel Hill. Children will also receive a thorough assessment of their behavioral development and a complete physical examination. Children will be invited back for similar follow-up assessments at 6, 12, 24, and 48 months of age. Children over two weeks of age may still be eligible to participate.
Contact: Danielle Spiker at (919) 843-3608 or danielle_spiker@med.unc.edu

DICER1- related Pleuropulmonary Blastoma Cancer Predisposition Syndrome: A Natural History Study

Pleuropulmonary blastoma is a rare lung tumor associated with the *DICER1*-related inherited cancer predisposition syndrome. Additional *DICER1*-related rare benign and malignant tumors include cystic nephroma, nasal chondromesenchymal hamartoma, ovarian Sertoli-Leydig cell tumor, ocular medulloepithelioma, and multinodular goiter. This study aims to further define the *DICER1* phenotype, evaluate the cancer risk, understand the *DICER1*-related syndrome biology, and identify other genes associated with these disorders. The study is being conducted by the Clinical Genetics Branch of the National Cancer Institute in collaboration with the International Pleuropulmonary Blastoma Registry, Children's National Medical Center, and Washington University. See website for enrollment and participation details:
http://dceg.cancer.gov/cgb/research/dicer1-ppb-study
Contact: Stephanie Steinbart, Referral Nurse, at (800) 518-8474

Genetic Causes of Ocular Conditions

The Medical College of Wisconsin is seeking individuals and families affected with ocular conditions including Peters Plus syndrome, Axenfeld-Rieger anomaly and syndrome, anophthalmia/microphthalmia, Peters anomaly, anterior segment dysgenesis (any), glaucoma, cataract, optic nerve hypoplasia/atrophy, and high myopia. Genes screened include *PITX2, PITX3, FOXE3, B3GALTL, CYP1B1, BMP4, SOX2, OTX2, VSX2,* and other candidate genes as they are discovered. Participation involves a blood or saliva sample and completion of a clinical/family history questionnaire.
Contact: Linda Reis, MS, CGC, Program Coordinator, (414) 955-7645, lreis@mcw.edu

Please send Research Network items to emily.place@gmail.com