Perspectives in Genetic Counseling
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President's Beat

NSGC Liaisons

Have you ever received an email or notice announcing an exciting genetics conference and thought, "Is anyone from the NSGC attending that meeting? Someone really should…" or, "Does this group know about genetic counselors? We should educate them about our expertise…" As genetics continues to be recognized as an integral part of all medical disciplines, new conferences, meetings, and workshops are being created to inform and educate a wide variety of audiences (such as healthcare providers, legislators, educators, the general public) about the genetic aspects of various diseases. These educational efforts may also impact and influence public policy.

As experts in the science of medical genetics and in communicating the meaning of that science, providing education, analyzing, interpreting and applying genetics knowledge, genetic counselors are in a unique position to add much to these meetings. In order to represent our profession and our society's vision, mission and strategic initiatives, the NSGC has several official liaisons. Liaisons are NSGC members who officially represent us to key organizations, national advisory committees, and initiatives.

The NSGC's liaison relationships are focused on organizations that are critical to our strategic objectives. An organization's priorities and goals must be in alignment with our mission, vision, and strategic plan – or must have an impact on our profession, now or in the future. NSGC liaisons attend important meetings to convey key NSGC messages, present and educate about our expertise, and identify important contacts, organizations, and/or activities in which the NSGC's initiatives and interests should be represented and involved. Liaisons provide summaries of their activities and communications with the liaison organizations, highlighting issues that are important to the NSGC.

As liaison relationships provide a mechanism for the NSGC to have influence and make inroads and significant contributions to the field of genetics, communication with the liaison organization about shared initiatives, goals, and vision is extremely important. Liaisons also provide ongoing analyses of the strengths, weaknesses, opportunities, and challenges involved in a relationship with an organization, committee, or initiative in the context of how the organization impacts the NSGC and the genetic counseling

profession. They also communicate what the NSGC can expect to gain from the relationship; after all, it's a two-way street! I've highlighted a few of our active liaisons below:

- **Cathy Wicklund** is a Program Director at Northwestern University in Chicago, Illinois and Past President of the NSGC. She is the NSGC liaison to the Institute of Medicine (IOM) Roundtable. The IOM is the health arm of the National Academy of Sciences and provides unbiased and authoritative advice to decision makers and the public. Cathy is extremely active as a member of the IOM, representing the NSGC's interests on key issues of importance to our society and profession. She is the only genetic counselor member of the group and, as such, demonstrates and promotes the value of our diverse skill set, training, and expertise to Roundtable members and conference attendees. As an example of Cathy's contribution on behalf of the NSGC, she recently co-directed a workshop on Integrating Large-Scale Genomic Information into Clinical Practice.

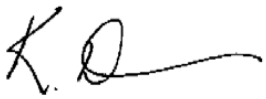
- **Scott Weissman** is an experienced cancer genetic counselor at the NorthShore University HealthSystem in Evanston, Illinois. Scott is the NSGC liaison to the American College of Surgeons Commission on Cancer and the National Accreditation Program for Breast Centers. He promotes the NSGC's key messages, including the importance of providing care to cancer patients from appropriately trained and qualified healthcare providers. Scott is involved in many activities with these groups, and they utilize his expertise in creating and updating standards of care for patients.

- **Cate Walsh-Vockley**, genetic counselor at the Children's Hospital of Pittsburgh in Pennsylvania, and **Michelle Fox**, genetic counselor at UCLA Medical Center in Los Angeles, California, are the NSGC liaisons to the Secretary's Advisory Committee on Heritable Disorders of Newborns and Children Education Task Force and Follow-up and Treatment Sub-Committee, respectively. They are active participants in the Sub-Committee meetings identifying potential public policy issues for the NSGC to follow, public health and newborn screening issues for genetic counselors, and areas in which additional genetic counselor participation and expertise could be utilized.

The NSGC Public Policy Committee oversees liaison appointments, as they are responsible for identifying and monitoring policy issues of interest to the NSGC. Through the Public Policy Committee and Executive Office, liaison activities are communicated back to the Board of Directors. As you can imagine, following and maintaining liaison relationships requires resources. To be sure this investment continues to provide benefits to you, our members, and remains aligned with our strategic plan, the Public Policy Committee and Board reassesses the utility of liaison relationships annually.

So the next time you see an advertisement for an upcoming, exciting genetics meeting, ask us if there is an NSGC liaison to that group! The NSGC's Board, Committees and

SIGs actively monitor genetics and healthcare organizations to determine whether input from the NSGC would help influence public policy or promote the value of genetic counselors to key audiences. If you have thoughts on a potential liaison relationship, please contact the Executive Office; your feedback will be provided to the Public Policy Committee and the NSGC's leadership.



Karin M. Dent, MS, LCGC
2011 NSGC President

The International Standards for Cytogenomic Arrays (ICSA) Consortium and its Genetic Counseling Workgroup Make Progress for Families and Genetic Counselors

By W. Andrew Faucett, MS, CGC; Director, Policy and Education, Genomic Medicine Institute at Geisinger Health System

Genetic counselors play a critical role in improving the clinical information available from genetic testing as it moves from research to clinical care. Recently, **Erin Riggs** (Emory University), **Karen Hanson** (Columbia University), **Melissa Savage** (Columbia University), **Karen Wain** (Mayo Medical Laboratories), **Darlene Ho Riethmaier** (GeneDx Laboratories), **Bethanny Smith-Packard** (Geisinger Health System) and **Andy Faucett** (Geisinger Health System), a genetic counseling workgroup, have been working with the International Standards for Cytogenomic Arrays (ISCA) Consortium (www.iscaconsortium.org) to improve the information available from chromosomal microarray (CMA) testing, in an effort to increase our ability to provide helpful information to patients and families seeking genetic counseling.

Laird Jackson, MD, has led the ISCA Consortium with a major initiative for clinicians to include clinical information when ordering CMA testing; as such, they have developed one-page phenotype submission forms for postnatal and prenatal CMA testing. The GC workgroup has also been concentrating on efforts to encourage genetic counselors, medical geneticists, and other clinicians ordering CMA testing to include clinical information.

Submitting clinical information on all patients at the time of testing improves the ability of clinical laboratories to evaluate CMA findings in terms of a patient's clinical presentation, and provide a more personalized result interpretation to improve genetic counseling. In addition, the more data that is submitted, the more robust the results database becomes, and the more information clinicians are able to provide back to families. As genetic counselors, it is important that we provide clinical information ourselves, and explain the importance of this to other healthcare providers. It is helpful to know that membership in ISCA is free and after registration, copies of the prenatal and postnatal phenotype forms can be downloaded from the ISCA website after clicking on "The Importance of Submitting Phenotypic Data".

The ISCA GC workgroup developed IMPACT (Initiative on Medical and Payor issues for Array-based Cytogenomic Testing) to improve third-party reimbursement for CMA testing in the postnatal setting. Recently, along with David Ledbetter, PhD, workgroup members met with the medical directors at the Geisinger Health Plan (GHP) and received approval of CMA (aCGH) testing for developmental delay. As of July 1, 2011, GHP policy states "array-based comparative genomic hybridization (aCGH) may be considered medically necessary when ordered by a Medical Geneticist, Certified Genetic Counselor, Pediatric Neurologist or Developmental Pediatrician." Not only did GHP approve CMA testing, it also recognized the important role that *genetic counselors* play

in the selection of appropriate genetic testing for our patients and families. The GHP policy currently provides coverage of postnatal evaluation of chromosomal imbalances in insured individuals who:

- "Exhibit congenital malformation(s), anomalies or dysmorphic features that are not specific to a well-delineated genetic syndrome; or
- Exhibit symptoms of non-syndromic developmental delay, intellectual disability or loss of developmental milestones; or
- Exhibit symptoms suspected of autism spectrum disorder"

The ISCA GC workgroup has posted example paragraphs collected from multiple genetic counselors, as well as a guide that can be used to develop "Letters of Medical Necessity" on the ISCA website under "Insurance Coverage Toolkit for CMA."

Working with David Miller, MD, PhD (Harvard University), the group distributed a survey ("IMPACT Survey Project") to genetic counselors to evaluate their experiences with insurance reimbursement for aCGH testing, and to learn more about the use of testing. Initial survey results from 153 genetic counselors show that over half of responding genetic counselors feel chromosomal microarray studies are indicated in greater than 25% of their patients, and about one third feel they are indicated in 5-25% of their patients.

Over 80% of survey participants have been involved in one or more cases where CMA test results directly impacted their patient's medical management, yet 85% of genetic counselors indicated they have had insurance deny coverage for CMA testing. The project is collecting clinical anecdotes that support changes in clinical management as a result of CMA testing results. A similar survey for medical geneticists and laboratory directors has been posted to the ISCA website, and a mailing to encourage participation will be sent to American College of Medical Genetics members in the near future.

Next steps for the ISCA GC workgroup include contacting insurers to discuss reimbursement for testing. The group has reviewed the policies posted on major insurers' websites about coverage of CMA, and many of the policy statements do not reference current publications or professional organization statements recommending CMA as the "first-tier diagnostic test." A list of these publications can be found in the ISCA website under "Insurance Coverage Toolkit for CMA."

The ISCA GC workgroup needs your help. Together, genetic counselors can increase the submission of clinical information with test requisitions, thereby increasing our ability to provide evidence-based genetic counseling to our patients. We also can work together to increase coverage of valuable genetic tests for our patients and families. To join in these efforts, contact the ISCA genetic counselors at isca@iscaconsortium.org.

Sarah Lawrence College Hosts Third Annual Genetic Counseling "Summer Camp"

By Sean Campbell, MFA, 2011, Sarah Lawrence College and Caroline Lieber, MS, CGC, Director, Joan H. Marks Graduate Program in Human Genetics

Despite recent advances in genetic medicine and the increasing need for individuals skilled in interpreting genetic information, the important role of genetic counselors and how they can enrich the lives of their patients is still widely unknown. Over the past decade, there has been incredible growth in the fields of genetics, and commensurate creation of new genetic counseling programs. However, many students interested in genetic counseling still know very little about this healthcare profession. Accessible literature on the field is sparse, and Internet searches yield only limited useful information.

Sarah Lawrence College, the first institution to offer a Master's degree program in human genetics and genetic counseling, has addressed this need for information with its Genetics Summer Camp for prospective genetic counselors. This year marked the third annual Genetics Summer Camp, which was held on June 6, 2011.

The campers, mostly undergraduate juniors and seniors – but also those looking to make a career change – have the opportunity to meet with professionals in the field and learn directly from their experiences. After a short breakfast, the campers are invited to sit in on a panel discussion providing an overview of genetic counseling and its role in modern healthcare. This is followed by a question-and-answer session between the campers and panelists.

Panel discussion at Sarah Lawrence College's 2011 Genetics Summer Camp

Following the panel discussion, campers are then treated to lunch and invited to participate in casual discussions with camp faculty regarding real-life case studies.

The next activity, the "Genetics Speed Sessions," have become a camper favorite. During these sessions, campers are broken into small groups and allowed to engage with genetic counselors in specialty areas including cancer genetics, prenatal genetics, and private industry. Discussions usually last for 15-20 minutes, at which point the groups rotate specialties, allowing contact with some of the concentrations the genetic counseling field has to offer. The camp then concludes with a wrap-up discussion, and interested campers are encouraged to stay in contact with camp faculty to pursue volunteer opportunities or internships.

Campers having discussions on the Sarah Lawrence College campus

Feedback from the Summer Camp has been astounding. Each year, we gain more attendees, furthering the dissemination of information about genetic counseling, and helping the future generation of genetic counselors make informed decisions about their career paths. This past year we had over forty campers in attendance, all of whom found the camp very informative. Many are looking forward to applying to genetic counseling programs. More can be said, but the campers themselves probably best describe their experiences from this year's camp:

"The number of different genetic counselors there was outstanding. It was great to meet so many different counselors with different jobs."

"So helpful for someone considering a career change, like me."

"Up the advertisement! This was such a great opportunity and I wish [that] I had [known] about [genetic counseling] earlier in my undergraduate career so I could explore the field more. It was great to see [so] many people younger than me interested in the field. I think the camp will definitely grow more in the future. Thank you again for the organization and efforts behind the camp!"

Suggestions for future programs include more case discussions, and perhaps some role-playing exercises. Overall, both the students and faculty left with a very positive feeling

about this kind of programming, and the faculty have all enthusiastically signed on to participate in the program again next year.

For Your Practice

National Hereditary Breast and Ovarian Cancer Week and National Previvor Day 2011

By Lisa Schlager, Facing Our Risk of Cancer Empowered (FORCE), Vice President, Community Affairs and Public Policy

In 2010, history was made when Facing Our Risk of Cancer Empowered (FORCE) worked with Congresswoman Debbie Wasserman Schultz (D-FL) to draft a resolution declaring the first-ever "National Hereditary Breast and Ovarian Cancer (HBOC) Week" and "National Previvor Day." HBOC Week marks the transition between National Ovarian Cancer Awareness Month (September) and National Breast Cancer Awareness Month (October), and highlights the genetic link between these two diseases.

National HBOC Week will take place **September 25 to October 2, 2011** and National Previvor Day is **September 28, 2011**. National HBOC Week and Previvor Day aim to raise awareness about hereditary cancer in communities throughout the U.S.

FORCE will be making its mission this week, and throughout the months of September and October 2011, to educate and empower those who are uninformed, and those who know about their hereditary predisposition to cancer – including women and men with *BRCA* mutations, anyone with a family history of cancer, breast and ovarian cancer survivors, and previvors (individuals who are living with a high risk of cancer but have not developed the disease).

To help genetic counselors, healthcare professionals, and others promote National HBOC Week and National Previvor Day in their practices and communities, FORCE has created an HBOC Week webpage, and the following resources:

- HBOC Week/Previvor Day 2011 poster with the warning signs of hereditary breast and ovarian cancer
- Video about hereditary breast and ovarian cancer
- HBOC Week/Previvor Day press release template
- List of books and movies about hereditary cancer

Events commemorating National HBOC Week and Previvor Day are planned throughout the country, including Passing of the Torch ceremonies with the Major League Baseball teams in Cincinnati and Washington, D.C.; a Major League Soccer game with the Philadelphia Union; a Previvor Day Art Exhibit in Phoenix; "In the Family" screenings; educational programs, and more. Many of these events are bringing together numerous groups in the breast and ovarian cancer communities. A list of scheduled events is also available on the website.

Although National HBOC Week focuses on *BRCA1* and *BRCA2*, it would also serve to call attention to other hereditary cancer syndromes.

"Awareness of an inherited predisposition to cancer may lead to earlier detection and preventive strategies that ultimately reduce the chance of dying from cancer. This is why I introduced House Resolution 1522," said Representative Wasserman Schultz. To see Debbie Wasserman Schultz's 2010 address to Congress, which preceded unanimous passage of the National HBOC Week resolution, visit: http://www.youtube.com/watch?v=9ocrQIyc8n0.

Licensure / Billing & Reimbursement

Coding Corner

Do You Need to Take Control of the Revenue You Generate? How to Become a Credentialed or Preferred Provider

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

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

We are happy to report that the 2011 online course *Take Control of the Revenue You Generate: How to Become a Credentialed or Preferred Provider* is now available for Continuing Education Units (CEU) credits. This course serves as a follow up to the 2009 course; *Learn the 3 C's to Maximize your Service Delivery Model: Coding, Credentialing and Compliance*. These courses aim to educate the NSGC membership on 1) the basic concepts and challenges of billing, 2) obtaining reimbursement for genetic counseling, and 3) ways to advocate for the profession.

After participating in *Take Control of the Revenue You Generate*, participants will be armed with the information needed to demonstrate that genetic counselors are uniquely qualified healthcare providers. Participants will be given the tools needed to strategically approach leaders within their institutions or local payers to promote recognition of genetic counselors as valuable providers. This means being the expert on the data supporting involvement of a trained and certified genetic counselor in a healthcare team, both in regards to health outcomes as well as to financial outcomes.

There are many facets to obtaining the highest level of fiscal success; genetic counselors need to take charge and be involved in the education of their institutional leaders and of their payer decision makers. For example, even if a genetic counselor is licensed, credentialing remains very important. Neither licensure nor credentialing alone guarantees reimbursement. This course will teach participants how to identify the various ways in which independent billable provider status can be obtained and the benefits and limitations of these.

Ultimately, no institution, state, or payer system will be identical, so each genetic counselor needs to be his/her own advocate and needs to be prepared to ask the right questions to determine the most applicable strategy to obtain professional recognition in their area. Becoming a successful, billable genetic counselor provider does not happen overnight – it will only occur if you take charge. We hope that this course provides you with the inspiration and background to get started!

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

SIG Speak

From the Health Information Technology Special Interest Group

Technology: The Good, the Bad, and the Necessary

By Heather Sellers, MS

Technology has changed the way we interact with others and with our world. In ten minutes, you can find the restaurant with the best pizza in town, text your friend to meet for a slice of pizza, look up directions to the pizza shop, reserve a table, and watch a video of a swimming dog biting a shark off the coast of Australia while you wait for your friend. Computers and smartphones have not just impacted entertainment, but they have begun to reshape the way health care is provided. A recent study by Fox et al, found that 61% of Americans use the Internet to search for health information and engage in other health-related activities; this group of individuals have been referred to as the "e-patient" (2009).[1]

The health care field has utilized technology to improve efficiency, cost effectiveness, and quality of care. The U.S. Department of Health and Human Services has worked to expand the use of electronic health records (EHR) and standardized electronic processes to reduce health disparities within the United States. Health information technology (HIT), like the medical record software Epic, has improved coordination of care, workflow efficiency, and use of medical teams. Due to this, the field of genetic counseling has an opportunity to further grow and expand through the use of technology.

Many genetic counselors have already embraced HIT and are forging development of new HIT to improve genetic counseling practices. At the last National Society of Genetic Counselors' Annual Education Conference (AEC) in Dallas, there were several posters and presentations on electronic risk assessment tools, red flag questionnaires, family health history questionnaires, and other tools intended to improve clinical efficiency and/or standardize practices. Many genetic counselors reported being asked to assist with integrating clinical genetics practice elements (informed consent documents, test results, pedigrees) into their new or existing EHR system. Subsequently, it was noted that there was no central place for experiences, ideas, and questions to be shared. The Health Information Technology (IT) Special Interest Group (SIG) was created to serve as a centralized resource for adoption, integration, and development of HIT.

The Health IT SIG hopes to accelerate HIT integration into the genetic counseling process. Members of the Health IT SIG will be able to participate in a discussion forum with genetics professionals at all stages of HIT engagement, gain efficiency from shared knowledge, and develop a more thorough understanding of the components and resources necessary for the successful implementation of HIT products and services. At the NSGC

AEC in San Diego this year, the Health IT and Familial Cancer Risk Counseling SIGs are co-sponsoring a session about the integration of HIT into cancer genetics. Four different genetic practices will share their experiences with developing and using HIT to standardize practices and improve efficiency. While originally developed for use in cancer genetics, the basic principles and ideas can be expanded into other areas like prenatal, pediatric, and adult genetics.

HIT will continue to grow and impact the provision of health care, so it is important for genetic counselors to step up and shape the future. Genetic counselors are often looking for ways to expand their practices, increase efficiency, and provide better services and care; technology is a tool that can be used to address all those issues. Perhaps in the future, a patient will have their entire family history (perhaps even entire genome) downloaded, be flagged as high risk, and be scheduled for an online risk assessment with a genetic counselor in as little as ten minutes!

If you would like more information about the Health IT SIG or are interested in joining, please e-mail co-chairs Megan Doerr at doerrm@ccf.org or Heather Sellers at heather.sellers@utsouthwestern.edu.

Reference:

1. The Social Life of Health Information. Accessed on August 5, 2011 at www.pewinternet.org/Reports/2009/8-The-Social-Life-of-Health-Information/01-Summary-of-Findings.aspx

NSGC News

Jane Engelberg Memorial Fellowship Student Research Award

By Christina Palmer, MS, PhD, CGC, Chair, JEMF Advisory Group

Since 1993, the Jane Engelberg Memorial Fellowship (JEMF) has been available to full members of the National Society of Genetic Counselors (NSGC) as a mechanism to obtain funds to pursue professional development and scholarly investigation related to the genetic counseling profession. The JEMF, established by the Engelberg Foundation in 1991 as an award administered through the NSGC, is an ongoing tribute to Jane Engelberg, who graduated from Sarah Lawrence College in 1973 with a Master's degree in Human Genetics.

2011 was a very exciting year for the JEMF Advisory Group (AG). For the first time in JEMF history, the AG solicited research proposals from genetic counseling students for the newly established **JEMF Student Research Award**.

Most genetic counseling training programs require a form of scholarly investigation, e.g., the thesis or capstone project. However, up until now opportunities for genetic counseling students to obtain funds to conduct their research have been limited. The AG – whose mission is to foster initiatives to improve the practice of genetic counseling – decided to expand its funding opportunities by creating an annual JEMF Student Research Award. The purpose of the JEMF Student Research Award is to foster research and grant writing skills at this early stage in students' genetic counseling training, which can continue to be used and honed throughout their careers.

The inaugural call for student research proposals was announced in March 2011, and by the June 8 deadline, 34 applications from 17 genetic counseling training programs representing both the United States and Canada had been received. Proposals were de-identified and assigned to AG members for review without institutional conflict by the NSGC Executive Office. The proposed research topics were varied, interesting, and timely. Proposals were evaluated using the following categories: Specific Aims and Objectives, Methodology, Feasibility (e.g., setting, timeline, or institutional support), Budget, Innovation, and Reviewer Enthusiasm (low, medium, high).

Due to the generally high quality of proposals the AG decided to fund six proposals at up to $500 each, instead of the maximum of five proposals they originally planned to fund. The awarded proposals were developed by students at six different training programs, and cover such topics as training program curriculum, genetic counseling practices, and family communication (see below for a description of each awarded proposal). Please join the AG in congratulating the JEMF Student Research Awardees at the JEMF session

during the 2011 Annual Education Conference in San Diego on **Saturday October 29, 2011, from 6-7 p.m.**

The JEMF Student Research Award was such a successful initiative that the AG will fund up to ten student research proposals in 2012 (maximum of $500 each). The AG is proud to continue Jane Engelberg's legacy through the JEMF Student Research Award.

Nuts and Bolts about the JEMF Student Research Award

- Eligibility: Students enrolled in accredited (or provisionally accredited) genetic counseling training programs

- Amount: maximum of $500. The JEMF AG philosophy is to award the best proposals, regardless of other funds that may be available for the research project.

- Budget: A budget must be submitted to justify the amount of requested funds. The award money can only be used for reimbursable items. Examples of reimbursable items include (but are not limited to) photocopying, survey software, transcription services, travel to sites for data collection, project-related parking fees, food for subjects, or subject reimbursements for items such as parking or an incentive. Requests for consulting fees for statistical or other support are acceptable. The award **cannot** be used for student, staff, or faculty salary/stipend. Awardees will submit receipts for approved budget items for reimbursement.

- Deadline: June 8 (if weekend or holiday, then the next business day). A call for proposals will be announced in advance, and will be posted to the JEMF website and the "Student Corner" section on the NSGC website.

2011 JEMF Student Research Award Winners

1. Katie Armstrong (University of Cincinnati)

Title: Perceived risk, worry, and satisfaction related to genetic counseling of childhood cancer survivors

With improvements in treatment strategies and better supportive care, the majority of children who are diagnosed with cancer are surviving well into adulthood. This growing population of childhood cancer survivors has led to the development of a new specialized area of healthcare – cancer survivorship care – focused on detection, provision of care, and counseling for the survivor regarding the late effects of therapy. Over the past thirty years, cancer survivor clinics have been developed that integrate a multidisciplinary model of care. Beginning in May 2007, the Cincinnati Children's Hospital Medical Center (CCHMC) Cancer Survivor Center added genetic counseling services to this comprehensive team of providers. In order to measure if these services have been

perceived to be valuable by patients and their parent/caregiver, two groups will be surveyed and compared in this study: those who have seen a genetic counselor in the Cancer Survivor Center (N=325) and those who have not seen a genetic counselor in the clinic (N=541). Questions will be asked to assess differences in perceived risk of cancer development and the level of worry related to cancer development in the cancer survivor and other family members, as well as satisfaction with genetic counseling among the group seen by a genetic counselor.

2. Brittany Batte (University of Michigan)

Title: Utilizing illness representations to improve family communication in a population at risk for cardiomyopathy

Communication of genetic risk, an important component of the genetic counseling process, is a family affair. The goal of our study is to understand how, what, when, where, and why information is communicated to at-risk family members, in order to ensure accurate medical management and improve health behaviors. We plan to explore the impact of individuals' illness representations on risk communication within their families in a hypertrophic cardiomyopathy (HCM) patient population. Illness representations, beliefs that people have about the cause of their disease, include perceived risk (perceived lifetime vulnerability to disease, including both likelihood and severity), perceived control (the power to influence their disease), and causal attributions, or "why me" (locus of control, controllability, and stability). We will assess the relationships between genetic test results (mutation positive and negative), illness representations, and family communication. We will also evaluate the impact of clinical severity, demographics, coping style, and uncertainty (perceived barriers and benefits). A better understanding of the factors that impact risk communication to family members of patients with HCM will help improve family communication overall. This has the potential to improve medical management for at-risk relatives, as well as reduce the incidence of sudden cardiac death associated with HCM.

3. Jessica Profato (Stanford University)

Title: Assessing the integration of genomics into genetic counseling training programs

Successful implementation of genomic medicine into clinical practice, including the incorporation of single nucleotide polymorphism (SNP)-based risk assessment, pharmacogenomics, and broad reaching results from exome/genome sequencing, will require health care professionals, especially genetic counselors, to be knowledgeable about the interpretation and limitations of genomic data and the approaches to providing genomic counseling. Some genetic counseling training programs may already be integrating these topics into their curricula, but this has not yet been studied. The goal of this project is to assess the extent to which genomic medicine, including genomics technologies, complex disease genetics, pharmacogenomics, and genomic counseling, is

being integrated into the curriculum of genetic counseling training programs, the ways in which it is being integrated, and the factors that facilitate or slow this integration. The project will involve surveying and interviewing program directors of American Board of Genetic Counseling accredited genetic counseling training programs, as well as a content analysis of curricular materials.

4. Jennifer Semotok (University of Toronto)

Title: Telling the children: Disclosure challenges and support needs of parents with children at risk for Huntington disease

Huntington disease (HD) is an autosomal dominant, progressive neurodegenerative condition with no known cure. A parent receiving a positive genetic test result for HD, along with their spouse/partner, faces the dilemma of telling their children about HD and their 50% risk for developing the same condition. Using a nationwide cross-sectional survey, we will sample a population of parents to examine their experiences with talking to their children about HD. This is the first large-scale HD study to identify the obstacles parents encounter when making decisions about disclosure to children and with the content of the disclosure process itself. Additionally, we will determine what support resources are beneficial and where gaps remain. This study expects to capture differences in the perceived challenges and support needs between parents who have tested gene positive for HD and their spouse/partners, as well as between parents who have disclosed and not disclosed. By identifying challenges experienced by parents related talking to their children about HD, this will provide invaluable insights into what role specific barriers play in decision-making and in the disclosure process. The data derived from this study will guide the development of resources to reduce or eliminate these barriers in the future.

5. Krista Sondergaard (Case Western Reserve University)

Title: Non-vascular Ehlers-Danlos syndrome and pregnancy

Ehlers-Danlos syndrome (EDS) is a group of heritable connective tissue disorders with genotypic and phenotypic heterogeneity. EDS, due to defects in collagen synthesis or collagen modification, has six subtypes, each with their own diagnostic criteria. Major clinical features of EDS include skin hyperextensibility, joint hypermobility, and tissue fragility. Little information is known about the effects of EDS on pregnancy. What information is available is generally regarding vascular EDS due to the increased risk for maternal morbidity and mortality during pregnancy. The purpose of this study is to elucidate the obstetrical experience of women with a non-vascular form of EDS. The researcher will survey women who have a clinical diagnosis of non-vascular EDS and have had at least one pregnancy about their obstetrical histories and what information they were given about the risks of pregnancy and EDS. Participants will be recruited at the Ehlers-Danlos National Foundation (EDNF) national meeting and by posting

advertisements for the survey at the EDNF website and in their monthly newsletter. It is hoped that the findings of this study will provide genetic counselors with specific risk assessments regarding EDS and pregnancy, and therefore genetic counselors will be more helpful to these women in providing anticipatory guidance.

6. Erica Wellington (Brandeis University)

Title: Cystic fibrosis carrier screening: Current practices and challenges in genetic counseling

Cystic fibrosis (CF) is one of the most common recessive genetic conditions in the Caucasian population, making it a frequent counseling topic in the prenatal clinic. However, because CF displays allelic and phenotypic heterogeneity, genetic counseling of carrier couples is often a challenging task. Depending on the mutations found, carrier couples are at risk of having a child anywhere along a phenotypic spectrum that includes classic CF, nonclassic CF, congenital bilateral absence of the vas deferens (CBAVD) and subclinical manifestations. Genotype-phenotype correlations are highly variable, and complex alleles, the effects of which are mediated by chromosomal background, further complicate counseling. Although there is a large body of literature describing the prognostic ambiguities associated with CF, the CF carrier screening and counseling practices of genetic counselors have not yet been described. This study will use an online survey and telephone interviews to evaluate prenatal genetic counselors' knowledge of CF carrier screening guidelines, describe current practices, and identify the genetic counseling challenges presented by complex screening scenarios. Establishing this baseline description of practices and challenges is one of the first steps toward helping genetic counselors provide accurate, consistent and useful CF counseling as part of quality preconception and prenatal patient care.

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With a Little Help From My (NSGC) Friends: My Experience with the NSGC Mentor Program

By Matthew L. Tschirgi, MS, CGC

I have been mentored in many different areas in my life, both personally and professionally. My experiences have always been positive; there is something energizing about learning from someone who is willing to share his/her experiences so I can improve myself. I have also been a mentor in different capacities, and have found it very satisfying to pass on my knowledge to those who are less experienced and are willing to learn from my victories and failures.

My decision to sign up for the National Society of Genetic Counselors' (NSGC) Mentor Program came after I graduated. I had been a genetic counselor for about two years, working for a private company since graduation. I thoroughly enjoyed meeting with patients, but had a growing interest in the business side of genetic counseling. When I first became a genetic counselor, I did not anticipate I would have such a strong interest in this area. However, I now know that I would like my career to eventually transition from a clinical setting to a corporate one.

I began to have questions: How would I accomplish this? What time frame do/should I have? Should I return to business school for formal training? What opportunities are there for genetic counselors interested in a corporate career? It seemed the questions flowed freely, but answers were elusive. So when I heard about the NSGC Mentor Program, I jumped at the opportunity right away. At this point, the NSGC Mentor Program was able to fill the gaps.

I signed up for the NSGC Mentor Program under the category "corporate and business practices." A few days later, I formed a match with a genetic counselor with thirteen years experience, who recently opened her own practice. Although I do not anticipate opening a private practice, I was excited to see what I could learn from her.

It has been very valuable to discuss my ideas from time to time with my mentor. Although she is two time zones away, this has not affected the mentoring experience. And while the official mentoring phase ended in the summer, my mentor and I continue to have a working relationship. I appreciate someone who can give her thoughts and ideas on my career path.

I've looked at several different options to transition into a corporate career, and it's been extremely beneficial to get my mentor's opinions. For example, she has given me valuable insights about how to navigate the "corporate" side of things, topics that interest me but did not come up during my genetic counseling training. These have included tips on marketing myself, advertising, forging relationships with potential clients and customers, and running a business. It may take five, ten or twenty years to complete my career goals, but I have no doubt that the NSGC Mentor Program will have aided me in accomplishing them.

I highly recommend the NSGC Mentor Program. While every mentor-mentee relationship will be different and the goals of the relationship will be different, if both the

mentor and mentee are willing to put a little time and investment into the relationship, the return can be tremendous.

Are you willing to put a little time and investment into a mentor-mentee relationship to see how you could benefit as either a mentor or mentee? Visit www.nsgcmentor.org to sign up for the Mentor Program. The next Match Phase is in ***November 2011****. Mentors can register between* ***November 1-15****; mentees can register and matches can be formed* ***November 16-30****.*

To join the NSGC Mentor Program, please visit www.nsgcmentor.org.

ABGC Update

Serving our Diplomates: Updates from the ABGC Credentials and Accreditation Committees

By the ABGC Board of Directors

Credentials

The American Board of Genetic Counseling (ABGC) wants to be sure that it is responsive to the needs of our diplomates. To this end, we recently conducted two surveys; our second Practice Analysis and a survey of recent and pending examinees to help the ABGC prioritize future opportunities related to the exam. We'd like to share some of the results of those surveys with you here.

The 2011 Practice Analysis Survey

The ABGC conducted its second Practice Analysis Survey in the first quarter of 2011. A detailed summary of the outcomes will be published in the Journal of Genetic Counseling in the near future. However, we'd like to share a few highlights. Most exciting was the response rate for the survey – an amazing 35% of genetic counselors participated. This is truly a remarkable response rate, since a typical response rate for this type of survey is in the range of 20-25%. Thank you for your dedication.

The primary purpose of a Practice Analysis is to derive the tasks, knowledge and abilities required in the daily activities of a genetic counselor. The compilation and analysis of the data from this analysis results in the creation of the Certification examination content outline. The newly derived content outline is not significantly different from the outline generated by the last Practice Analysis in 2008; however, one new major sub-content area was created: Communication. This new area consolidates tasks from the 2008 content outline and supports our ability to ensure adequate testing of knowledge, skills and abilities in this area. The ABGC will be releasing the examination content outline later this Fall, along with information on when the examination will reflect these changes. At this time, we estimate that the new content outline will be implemented with the 2012 Certification examination. Look for the announcement of the new examination content outline on our website (www.abgc.net) later this year.

Survey of 2010 examinees and current genetic counseling students – enhancing the Certification examination

The ABGC recently surveyed the newest members of the profession regarding their opinions on the prioritization of five possible enhancements to the Certification examination. Current genetic counseling students, 2011 program graduates, and those who took the exam in 2010 were eligible to complete the survey. We received responses from 439 diplomates and trainees.

Overwhelmingly, a practice examination was ranked most valuable (69%). This was followed by a self-assessment examination, instant scoring, and having more than one test window. Moving the test window was perceived as least valuable and the majority of respondents (~70%) indicated that they do not have a preference for the month in which the examination is offered. The ABGC plans to eventually implement most of these improvements and is currently working on developing a practice examination.

Accreditation

The ABGC has formed a separate taskforce whose charge is to develop and implement a business plan that allows the organization to separate its two core businesses, Credentialing and Accreditation, into two separate board entities. This Transition Taskforce meets monthly, and is currently working towards separation in 2013. To prepare for this separation, the ABGC has undertaken a number of initiatives to ensure all processes and policies are in order for the transition.

The ABGC is also in the process of reviewing the "Required Criteria for Graduate Programs in Genetic Counseling." This process is aimed at reducing redundancies, while increasing comprehension and readability to ensure adherence to the standards. The taskforce charged with completing this initiative is ahead of schedule, and a new document and application form should be available by late Fall.

Finally, the ABGC has asked thought leaders in our field to help review and update, as needed, the Practice Based Competencies (PBC). The PBCs define the minimum skill set required of newly graduating genetic counselors and help inform multiple aspects of graduate training. This group will meet October 4-5, 2011 and a final report will be generated following the meeting.

In other news, there are multiple new genetic counseling graduate programs in the accreditation pipeline. Emory University in Atlanta, Georgia has been granted provisional accreditation. The ABGC is expecting four programs to apply for full accreditation in 2012. Pending Board approval of all accreditation applications and with the addition of Emory, that will bring the total number of ABGC-accredited programs to thirty-three.

The Board of Directors encourages you to attend the ABGC Business Meeting scheduled during the NSGC Annual Education Conference in San Diego, California on **Sunday, October 30** for the latest updates.

Student Forum

GenetAssist launches pilot program in Guatemala

By Ny Hoang, MS, Gillian Blaber, MS, and Lindsey Alico, MS, Joan H. Marks Graduate Program in Human Genetics at Sarah Lawrence College, Class of 2011

The GenetAssist Team (from left): Caroline Lieber, Gillian Blaber, Dr. Marta Julia Ruiz, Lindsey Alico, and Ny Hoang

GenetAssist was created in the fall of 2010 with the goal of promoting global access to genetic counseling services. Three students attending the Joan H. Marks Graduate Program in Human Genetics at Sarah Lawrence College, **Lindsey Alico**, **Gillian Blaber**, and **Ny Hoang**, founded *GenetAssist* along with our program director, **Caroline Lieber**. The team envisioned a global outreach organization of trained and culturally sensitive genetic counselors that could travel to countries in need of genetic services to assist with genetic education and service provision. To tackle this large undertaking, three separate capstone projects were created, each focusing on a different component of the organization. The team met regularly to collaborate and correspond with potential clients, including doctors and other healthcare providers interested in establishing or improving genetic services in their country.

GenetAssist's vision gained momentum when Dr. Marta Julia Ruiz, a native of Antigua, Guatemala, reached out to Lieber during a visit to Sarah Lawrence College. Dr. Ruiz traveled to the United States to receive an award for her work on the Population Council's *Abriendo Oportunidades* (Opening Opportunities) project, which strives to provide "opportunities for Mayan girls and young women."[1] Dr. Ruiz explained that she had recently cared for an infant who died of a congenital anomaly immediately after birth, and although she suspected a genetic etiology, an official diagnosis was never made. Dr. Ruiz expressed her concern about the lack of genetic knowledge and services in Guatemala, and her desire to provide education to health care professionals and community members. It seemed like a perfect opportunity for both parties. By December

[1] http://www.popcouncil.org/projects/244_CreateOpportunitiesMayan.asp

2010, *GenetAssist* had funding in place through an anonymous donor and the team began preparations to travel to Guatemala and launch a pilot program.

In March 2011, the four of us traveled to Antigua, Guatemala for a weeklong visit to accomplish our first goal: to perform a genetic service needs assessment. During our stay, the *GenetAssist* team had the chance to meet with many medical professionals at public and private hospitals in Guatemala City, Antigua, and Solola. Each meeting introduced us to new knowledge and insights regarding the health care system in Guatemala. Particularly, this informed us about the role, if any, that genetics currently plays and the potential for what it could in the future. The team learned that the conditions of greatest concern are neural tube defects, cleft lip and palate, chromosomal abnormalities, and metabolic diseases. Many of these conditions are seen at an increased rate in Guatemala, yet the numbers of doctors able to detect or treat them are limited, as formal training in genetics is not available in the country's medical schools. Additionally, genetic testing is often both unavailable and costly, leaving doctors limited to clinical diagnoses.

One topic broached in every meeting was the lack of genetic knowledge and education among both the general and medical population. The *GenetAssist* team learned that some populations in Guatemala hold on to traditional beliefs as the explanations for medical problems. For example, one popular belief is that 'bad karma' can cause birth defects and genetic conditions in children. Another links using prenatal vitamins and folic acid with adverse pregnancy outcomes. Drawing from this experience, *GenetAssist* hopes to help increase genetic literacy in Guatemala and dispel myths and misconceptions about the etiology of genetic conditions by increasing access to genetic information, while still acknowledging and respecting the cultural beliefs of the area.

From our survey of the medical professionals we encountered, we learned that the majority had little to no genetic education, and were adamant about the need for this type of training. They felt that genetic education is especially important for midwives, who care for more than 80% of the nation's pregnancies, often without formal training. The medical community was also insistent that although there are many pressing health care issues in Guatemala such as infectious diseases, clinical genetic services are important and should not be discounted. Each meeting brought echoes of the notable lack of genetic services and testing available to these communities, and an undeniable need and desire for them.

This trip was an important first step in determining the need for genetic services and the availability of technology and resources in Guatemala. The next step will be to devise a strategic plan for successfully implementing and sustaining genetic education and services in Guatemala, given the economic and cultural constraints. *GenetAssist* is currently exploring different funding sources to support these initiatives. Because of its affiliation with Sarah Lawrence College, *GenetAssist* will have an endless reserve of genetic counseling students to continue working in Guatemala and to plan future collaborations with other countries. **Zoë Nelson**, a second year student in the Human Genetics Program at Sarah Lawrence College, has recently joined the *GenetAssist* team. For her capstone project she will be developing an educational module in basic science

and genetics that can be incorporated into Dr. Ruiz's *Abriendo Oportunidades* program. Through the continued efforts of the *GenetAssist* team, we hope to be able to provide access to genetic information, education and services wherever they are needed.

For more information on *GenetAssist*, please visit our team blog at:
http://genetassist.blogspot.com/

The New Graduate Life

From Student to Supervisor:
My Experience Working Where I Went to School

By Amber Mathiesen, MS, LCGC, University of Utah, Class of 2009

My experience with the University of Utah's Graduate Program in Genetic Counseling began the day I shadowed a genetic counselor. I remember walking into the hospital and going up the elevator to the office. I felt excited and nervous because I knew this was what I wanted to do with my life, and I was now taking the steps to make my life go in that direction. It was a great day; I observed patients and spoke with a genetic counselor who told me about his role with patients and working with graduate students.

As I went to leave, the elevator doors closing behind me, I thought to myself, "Wow I would love to work in a place like this someday; in fact I would love to work *here*." Little did I know that it was exactly where I would go to graduate school, and where I would end up working in the career that I love.

As a student I often thought about how much I enjoyed the program, and dreamt that one day I would be working as a part of it. So you can imagine my delight when I was offered a position to do so. Here I am, a University of Utah graduate, practicing as a genetic counselor and participating in the graduate program as a clinical supervisor, guest lecturer, and mentor.

However, I must admit that I was initially nervous. I felt this way because I would soon be working with the people who knew me as a student and whom I knew as supervisors, mentors, and teachers. These were the people who had been with me through it all; they witnessed me ecstatic with accomplishment and weary with defeat. They knew my strengths and weaknesses. They witnessed my growth as a genetic counseling student and everything that came with it – the good, the bad, and the ugly. Furthermore, these were the people I looked up to, sought advice from, and admired. Despite this, I knew I was

going to need to grow and be someone bigger than I had previously known myself to be. I was no longer a student – I was a genetic counseling professional.

This career path not only modified how I relate to the people around me and to myself, but also provided an opportunity for me to witness an experience from different viewpoints. I have seen the graduate program from "both sides" of the fence. On one side I am an alumna to the program; this is exciting because it allows me to help the current students in an effective, accessible way. I have passed the program's courses, completed the research requirements, and gone through the rotations. This helps me speak to students and answer their questions.

Students also often share their excitement and worry about their current course loads, progress in their research projects, and development of skills in their rotations. In addition to listening and giving advice, I also relate to what they talk about and remember my own experiences in similar situations. This has allowed me to stay connected to what it was like to be a student, which has contributed to my professional development as a supervisor and mentor. This has been important during this process of development because it requires me to participate in self reflection, which from my experience, results in growth. Furthermore, I believe staying connected allows me to listen to students with empathy and understanding.

On the other side, I am now a working professional – a supervisor, mentor, and lecturer. I can now say there is significant truth in the statement, "If you want to master something, teach it." Every day I interact with a student, I learn something new. As a relatively new graduate I find this both challenging and exhilarating. I've had to be patient with my own learning process through this time, realizing that I, too, am still learning.

From the particular vantage point of working where I went to school, I have also realized how, as a student, I was often oblivious to the logistics of the program. I remember my first program meeting as a professional, thinking about how much actually went on behind the scenes to make the program work. Additionally, I was unaware of the degree of commitment and time people spent to help me become the genetic counselor I am today. This experience has opened my eyes and made me realize that things are not always what they seem, that there are often components to life that we do not see, and that there are many perspectives to a single situation.

It is always fun to reminisce and remember that day when I began my experience job shadowing. Looking back at this reference point makes me realize how far I've come, and how much I've learned. This is something we often don't think about in our day-to-day routines. It is also a good reminder that life takes us to great places, full of experiences that allow us to learn and develop into the people and professionals we always hoped we would become.

Genetic Counselor Publications

Feature Article

By Sara Spencer, MS, CGC

Facio FM, **Brooks S**, **Loewenstein J**, Green S, Biesecker LB, **Biesecker BB**. Motivators for participation in a whole-genome sequencing study: implications for translational genomics research. *Eur J Hum Genet*. 2011 Jul 6. [Epublication ahead of print]

Flavia Facio, MS, CGC

Whole-genome and whole-exome sequencing technology is here, and much sooner than had been expected. With the cost of full-genome sequencing now lower than, or comparable to, some other clinically available genetic tests, it behooves our field to explore the social behavior of individuals interested in having full-genome sequencing. **Flavia Facio, MS, CGC** and her colleagues at the National Institutes of Health (NIH) have positioned themselves at the vanguard of this task.

Flavia's undertakings are impressive. Until recently, she worked on a number of spin-off research projects related to her current study. Unlike many genetic counselors, her position was one hundred percent research, of which she spent approximately thirty percent specifically on genetic counseling research. She has been working at the NIH for about seven years and, in her current position as associate investigator, she is devoting her time to completing a number of projects relevant to the practice of genetic counseling.

Flavia and her colleagues coordinate a study called ClinSeq®.[1] The study aims to enroll a cohort of more than 1,000 participants, ages 45 to 65, who consent to full-genome sequencing. Flavia's recent publication in *European Journal of Human Genetics* surveyed a subset of this cohort to assess their motivations and expectations of whole-genome sequencing. She states, "Genetic counselors are well positioned to provide counseling for individuals who avail themselves of this new technology. As genomic

sequencing technology becomes more widely used in research and enters clinical practice, it is important that we start to investigate the motivations and expectations of our clients in seeking their own genomic information. Our recent publication provides an initial glimpse into the motivations of early adopters of this new technology. This and other similar studies that seek the input of research participants and patients will provide a foundation for additional genetic counseling research, which can eventually guide our practice in the era genomic medicine."

The field of genetic counseling also needs to prepare itself for the interpretation and communication of this information to patients. With this technology on the clinical horizon and the anticipated departure from our traditional genotype-to-phenotype process of counseling, genetic counselors will need to understand what study participants and, ultimately, patients seek from full-genome sequencing. Flavia proposes this as another population to study, to build on her team's recent research.

Flavia has given out a few positive results from the ClinSeq® study thus far. She stresses that the potential for getting results that have profound effects for participants is a reality, since whole-genome and whole-exome sequencing analyze more than single nucleotide polymorphisms (SNPs) alone. Most genetic counselors faced with the task of delivering results from full-genome sequencing would first seek guidelines for when to, and when not to, disclose a result.

Luckily, Flavia and her colleagues are well ahead of the game here, too! A manuscript describing a few of these positive result cases, the algorithm or process that they followed to make these decisions, and their approaches for returning the information to the participants has been submitted for publication. Further research is underway at the NIH that explores attitudes and intentions of participants in the ClinSeq® cohort on learning different types of results from whole-genome or whole-exome sequencing, (e.g., carrier status or variants of uncertain clinical significance). The results of this study will provide guidance for when we as genetic counselors are faced with delivering these types of results in the near future.

Flavia has always enjoyed research and even sought out research projects during her graduate training in Human Genetics at Sarah Lawrence College. Flavia finds research very gratifying. She states, "What draws me to [research] is the process itself." She finds it interesting and stimulating to come up with research questions, find ways to answer them, analyze the results, and write the material up for publication. She adds that she enjoys the collaborative process and working with people in other disciplines. She also views research as an important opportunity to contribute to the field of genetic counseling, where additional research is much needed. Flavia would like to continue focusing on research for the rest of her career.

Flavia has advice for those genetic counselors or new graduates interested in contributing to research in our field. She offers, "Become familiar with, or make it a habit to read the literature so that you can identify… gaps" that could give rise to research questions. She

also recommends finding people with whom you can collaborate, and mentors to help with your research agenda.

Flavia plans to have preliminary results from her studies to present at the 2011 NSGC Annual Education Conference. She hopes to see you there!

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(Names of genetic counselors appear in bold)

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*Authors contributed equally to the publication

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

AEC Update

See You in San Diego at the 2011 Annual Education Conference!

By Elizabeth Wood Denne, MS, CGC, 2011 AEC Chair and
Claire N. Singletary, MS, CGC, 2011 AEC Vice-Chair

The AEC is turning 30! We are thrilled to invite you to join us in San Diego, California for the 30th Annual Education Conference (AEC). The AEC will be held at the waterfront San Diego Marriott Marquis & Marina, which is less than five miles from the San Diego International Airport (SAN).

San Diego Marriott Marquis & Marina
(www.marriott.com)

The world famous San Diego Zoo in Balboa Park is less than five miles from the Marriott, as is historic Old Town San Diego. The Seaport Village and the Gaslamp Quarter are both within walking distance of the Marriott and feature great restaurants and shopping. San Diego is famous for its waterfront beauty, nearby beaches, golf courses and sports; the Major League Baseball's Padres and the National Football League's Chargers both call San Diego home. You should have already received the preliminary program brochure with all of the dates and deadlines for the AEC, which will be held **October 27-30, 2011.** We look forward to celebrating our thirtieth year with you.

Pre-Conference Symposia

Are you looking for a more in-depth presentation and discussion about a favorite genetic topic? Don't forget to register for a Pre-Conference Symposium! The Pre-Conference

Symposia will take place on **Thursday, October 27**. The optional Pre-Conference Symposia will offer an opportunity to gain new perspectives and a deeper understanding of six cutting edge topics. Pre-Conference Symposia sessions include:

- 101 - Challenges of Extracting Clinically Relevant Data from Whole Genome and Exome Sequencing
- 102 - Down Syndrome: First Trimester to Birth
- 103 - Cancer News: Expert Views
- 104 - Diversity, Cultural Competence and Genetic Counseling
- 105 - Grant-writing Bootcamp: A Constructive, Intensive, Personal Session Geared Towards Helping You Secure Project Funding
- 106 - All Bones About It: Medical, Psychosocial and Legal Issues in Pre- and Postnatal Counseling for Skeletal Dysplasias

Each session will last five hours, allowing for a deeper review and discussion of the topics. We anticipate the attendance at each symposium will be smaller than at an Educational Breakout Session, which will allow for a more interactive experience. Each symposium will require registration separate from the AEC and will have limited space available, so sign up early.

AEC "Book Club"

We are excited to have authors Dina Roth Port and Bonnie J. Rough joining us for Plenary Session 203 - "Beyond the Page: Insight Into the Personal Impact of Carrying Gene Mutations" on **Friday, October 28**. Dina Roth Port is the author of "Previvors," which follows five healthy women who learn of their predisposition to breast and ovarian cancer through *BRCA* testing; this book was recently reviewed in the Journal of Genetic Counseling (June 2011).

Bonnie J. Rough is the author of "CARRIER: Untangling the Danger in My DNA," which is her own personal account of researching her family history of hypohidrotic ectodermal dysplasia and how this impacted her journey to motherhood. If interested, we encourage AEC attendees to read both books prior to the AEC. Both authors will also be selling and signing copies of their books following the plenary.

Sessions Available Online After the AEC

Once again, sessions from the 2011 AEC will be available online after the conference. In early 2012, a recording of the conference sessions along with synchronized PowerPoint presentations will be available. Register for access to the online sessions before the conference. For those of you who won't be able to join us on-site in San Diego, these recordings will be available for purchase to all members after the AEC and can be used to obtain CEUs.

Program Book: Print Your Notes Before the Meeting

To continue our effort to be "green," speakers' notes and PowerPoint presentations will be available online prior to the conference instead of in the onsite program book. The membership will be notified when the presentations are posted. We encourage you to decide which talks you cannot miss and print out the presentations you want to have on hand at the conference ahead of time. A suggestion: read over the presentations on the plane and spend your first night catching up with friends instead of flipping through a big program book!

The NSGC will offer an Internet pavilion, but only for viewing purposes as there will not be printers. If you need to print materials on-site, the hotel business center is available for a fee. If you bring your laptop or tablet computer, another option is to download the presentations ahead of time for viewing during the actual sessions (note: wireless internet is not available in the conference rooms and power cords will not be supplied). A smaller program book will be distributed at registration with the AEC schedule and hotel information so you will have something in hand to find your way around.

Networking, Networking, Networking

The AEC is not only a great educational opportunity but a prime networking opportunity. The Welcome Reception is always a good place to see colleagues and friends and should not be missed. Due to the busy agenda, we are fortunate to offer two networking receptions this year. Please join us for the Welcome Reception on **Thursday, October 27 from 6:15-8:15 p.m.** Additionally, there will be a sponsored reception for all AEC attendees sponsored by the Boulder Abortion Clinic on **Friday, October 28 from 5:30-7:00 p.m.** A list of registered attendees will be sent to all conference attendees before the meeting, so you can see who will be at the AEC and set up some additional time for networking activities before you arrive.

Outreach Event

In an effort to reach out to the community of our host city, the NSGC annually conducts an Outreach Event during the AEC. This year's event is being coordinated by **Debra Han**. Debra and her Outreach Subcommittee have been hard at work presenting a PowerPoint presentation entitled "Genetic Counseling as a Profession" to high school and college students in the San Diego area. The students who attend these presentations have been invited to join us for an afternoon during the AEC to attend educational sessions and to hear a panel discussion of genetic counselors from a variety of job experiences and work settings. Many thanks to Debra and the AEC Outreach Subcommittee for all of their hard work. We know that this year's outreach event will be a great success.

Abstract Update

Best Full Member Abstract Award: This award will include a monetary prize, as well as the opportunity to present the research in a plenary session. It will be presented to the Full Member who submitted the best abstract as judged by members of the Abstract Workgroup.

Best Student Abstract Award: This award will include a monetary prize, as well as the opportunity to present the research in a plenary session. It will be presented to the Student Member who submitted the best abstract as judged by members of the Abstract Workgroup.

Best Poster Award: This will be awarded after judging during the "Posters with Authors" session. The winner will be announced later in the conference and will receive a monetary prize.

Late-Breaking Session

Based on the success of the late-breaking session at the 2010 conference, we have reserved time on **Sunday, October 30** for a late-breaking topic. This year we are excited to have Dr. Wayne Grody, president of The American College of Medical Genetics (ACMG), as our speaker for this session. The topic of his presentation will be determined closer to the conference.

Register Now

It's not too late to register for the AEC! Join us in San Diego for a great educational opportunity, while catching up with old friends and meeting new colleagues. For more information, please visit the AEC webpage at http://www.nsgc.org/Education/2011AnnualEducationConference/tabid/356/Default.aspx

If you have questions, please contact **Elizabeth Wood Denne** (ewdenne@jhmi.edu) or **Claire N. Singletary** (Claire.n.singletary@uth.tmc.edu)

Resources / Book Review

Reviewed by C. Shai Huffard-King, MS, CGC

Positive Results: Making the Best Decisions When You're at High Risk for Breast or Ovarian Cancer
Author: Joi L. Morris and Ora K. Gordon, MD
Publisher: Prometheus Books
Pages: 395
Retail price: $20.00
ISBN: 978-1-59102-776-8

Joi Morris and Dr. Ora Gordon team up for Positive Results, a hereditary breast and ovarian cancer syndrome (HBOC) guidebook that provides facts about cancer risks and the various screening and risk-reducing options. In addition, Positive Results also walks us through Joi's *BRCA* journey, her quest for more information, and how she made many difficult decisions. The book has a very compassionate tone and addresses the gravity of harboring a *BRCA* mutation with the optimism necessary to cope. Despite the nature of the subject it is an enjoyable read, as it is peppered with Joi's personal experiences, the experiences of others, and glimpses into who they are and how they made their medical management decisions. I cannot think of a better partnership for a handbook like this: Joi, a *BRCA2* previvor and Facing Our Risk of Cancer Empowered (FORCE) outreach coordinator with a journalism degree, and Dr. Gordon, a compassionate and experienced medical geneticist – *genius*.

When the patient hears, "You have tested positive," it is the beginning of a lifelong journey of questions – some with answers – and potentially difficult decision making. Often times, these individuals race to the Internet to research their options, unaware of whether or not they can successfully sift through the good, the bad, and the ugly information on the Web. Some health care providers don't provide the whole picture and may sway patients towards one type of surgery or surveillance only. It is imperative that all high risk patients have a true understanding of the risks, benefits, and limitations of each appropriate screening modality, surgical intervention, and chemoprevention available. For example, they should be educated on the differences between skin-sparing and nipple-sparing mastectomies, implant versus autologous tissue reconstruction, and whether or not their uterus should be removed at the time of oophorectomy. Fortunately, Positive Results provides the reader with a clear, unbiased comparison of all possible HBOC surveillance recommendations, risk-reducing options, and surgical options, allowing patients to approach their healthcare team armed with a sense of empowerment as they plan to make potentially life-saving choices.

Positive Results has been described as one part memoir and three parts guidebook, and serves as an extension of genetic counseling. Part 1 provides a detailed and comprehensible introduction to genetics, the history of *BRCA1* and *BRCA2*, and the

importance of genetic counseling and risk assessment, and also encourages the patient to ask themselves, “Do I really want to know?”

Part 2 addresses hereditary cancer risk and provides concise summaries and valuable tables of numerous journal articles and prominent *BRCA* research. In fact, Dr. Gordon addresses the dearth of research on risk modifiers such as oral contraception, soy, green tea, and others. She especially addresses the healthy diet risk modifier with her “Two-Week, Breast-Health Blitz.”

Part 3 comprises half of the book, and details the medical management options for those at high risk for breast and ovarian cancer. Here, Joi shares her journey of choosing prophylactic bilateral mastectomies and delaying oophorectomy until her late forties. Her experience, as well as those of others, provides unique insights into how she came to her decision about her medical management and how she shared her surgery decision with her young sons. Joi’s story reminds us that these women and families need support and encouragement long after they leave our offices.

Positive Results is a valuable handbook for patients and their families at elevated risk for breast and ovarian cancer, as well as for the clinicians caring for them. It covers the a-to-z of *BRCA* and is a perfect quick reference manual for busy genetic counselors. Most importantly, this book provides comfort to the high risk reader, so they are not traveling the *BRCA* road alone.

Research Network

By Emily Place, MS, CGC

Study of Smith-Lemli-Opitz Syndrome

Oregon Health & Science University (OHSU) in Portland, Oregon is recruiting participants to take part in a natural history study of Smith-Lemli-Opitz Syndrome (SLOS). As part of this natural history study of SLOS, every year or every other year clinical and biochemical information is collected from advanced sterol tests, brain magnetic resonance imaging (MRI) and magnetic resonance spectrosocopy (MRS), hearing and vision evaluations, and extensive behavioral, developmental and feeding evaluations. OHSU is part of the Rare Diseases Clinical Research Network (RDCRN), a network of consortia across North America dedicated to clinical research and collaboration with patient advocacy groups for rare diseases such as SLOS. (For further information, visit the RDCRN website: http://rarediseasesnetwork.epi.usf.edu/). STAIR (for Sterol and Isoprenoid Disorders Research) consortium is focused on continuing research on SLOS and other rare sterol and isoprenoid disorders. OHSU's consortium also includes the following other sites: University of Nebraska Medical Center, Cincinnati Children Hospital Medical Center, Children's Hospital Pittsburgh/University of Pittsburgh Medical Center, and National Institutes of Health / National Institute of Child Health and Human Development Intramural Research Program.

Contact: Jessica Adsit, MS, CGC at 503-494-6524, adsit@ohsu.edu

Simons VIP Connect

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

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

The *PTEN* Study

The *PTEN* study at the Cleveland Clinic Foundation is actively recruiting patients with characteristics suspicious for a germline *PTEN* alteration and patients with known mutations. All samples undergo mutation scanning and promoter sequencing and select samples will undergo multiplex ligation-dependent probe amplification (MLPA). The referring provider will be notified whether results are negative or if a variant, single nucleotide polymorphism (SNP), or deleterious mutation is found. Nomenclature will be shared with the clinical lab of choice for site-specific confirmation of deleterious results. The Principal Investigator is Charis Eng, MD, PhD. For more information, visit the website http://www.lerner.ccf.org/gmi/research/pten/.

Contact: Jessica Mester, MS CGC at pten@ccf.org

Study of the mechanisms of chromosome rearrangements

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

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

Genetic Epidemiology of Pancreatic Cancer (PACGENE) Study

Researchers at Wayne State University, Mayo Clinic, Johns Hopkins University, MD Anderson Cancer Center, Dana Farber Cancer Institute, and University of Toronto aim to map one or more pancreatic cancer susceptibility genes. The study is currently enrolling families with at least two cases of pancreatic adenocarcinoma other than parent/child pairs. Participation includes phone interview or mailed questionnaire, medical record review, contacting family members, and donation of a blood (for those with cancer), archival tissue or saliva sample. Married-in family members can participate as part of a control group. Participants will be compensated $25-50 U.S. dollars for their time. Travel is not necessary. Families will not receive individual test results. Affected individuals need not be living; however, a DNA sample, such as tissue, must be available on at least one affected in families with three or more cases, or on *both* cases in families with only two affected. For more information, visit the website http://www.karmanos.org/cancer.asp?id=927&cid=19.

Contact: Kate Sargent, MS, CGC at 313-578-4240 or sargentk@med.wayne.edu.

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