Perspectives in Genetic Counseling
Volume 30, Number 2
Summer 2008

Jessica Mandell, MS, CGC
Editor
jmandell@slc.edu

President's Beat

NSGC recognizes the importance of increasing the diversity of the genetic counseling profession to help reduce health disparities in genetic counseling services. A critical step in achieving diversity is to make our organization welcoming to and inclusive of all potential members regardless of race, ethnicity, gender, sexual preference, religion, practice type, social group or other variables. This diversity requires NSGC leadership, staff and membership to become aware of, understand and respect their beliefs and values in relationship to others. NSGC also must help its leadership, staff and members develop and enhance skills to work with colleagues from under-represented groups at the organizational level - organizational cultural competence.

Work in Isolation

The NSGC and its members have taken a number of steps toward addressing diversity and cultural competence within the organization. Initially, some of these steps were member-driven (such as the development of the Diversity SIG and select initiatives of the Diversity Subcommittee). Most of these efforts were done in isolation, though, without a shared history of what had and had not been undertaken in the past.

This isolation began to change in 2005 when NSGC President, **Kelly Ormond**, identified diversity issues as one of her goals. To this end, Ormond held a retreat, facilitated by member, **Dr. Vivian Ota Wang**, to address racial and ethnic disparities in the NSGC membership and professional activities. The retreat revealed four key issues:

1) The makeup of the NSGC general membership and volunteer leadership had not substantially changed over time with respect to diversity.
2) Many NSGC members who self-identify with diverse racial and ethnic groups experience NSGC and the professional climate as insincere, with a majority of the general membership often imposing unsupported beliefs regarding assumptions based on assumed ethnic/racial status.
3) The organization continues to be challenged in creating an initial and enduring welcoming professional environment for all of its members.
4) More coordinated efforts are needed for recruiting individuals from a wider breadth of racial and ethnic diversity.

Developing Organizational Cultural Competence

As a result of this retreat and discussions afterward, the NSGC Board decided that a key step to increasing diversity was to strive for organizational cultural competence. This issue was considered of such import that it was included in the 2007-2009 strategic plan. In 2007, NSGC provided feedback on multiple bills addressing health disparities and increasing diversity in the workforce. Then, in 2008, Dr. Wang was invited to the February 2008 NSGC Board of Directors meeting to discuss how organizational cultural competence could be achieved.

The Board learned that one of the first steps is for leadership and staff to achieve the highest level of cultural competence possible to provide a nurturing, respectful, inclusive environment. This requires a commitment to ongoing cultural competence training. Other fundamental components of organizational cultural competence include:

- having a clearly articulated vision regarding the importance of diversity and inclusion
- conducting a climate survey to assess the degree to which members of various groups perceive opportunities available through the organization and the ways they feel valued, rewarded, and/or are disenfranchised
- providing ongoing education
- mentoring and evaluating NSGC staff and volunteer leaders to promote the expectations and skills necessary for developing a culturally competent organization, and
- establishing outcome measures to evaluate the success of such endeavors.

New Task Force at Work

To address these fundamental components, the NSGC Board has appointed an Organizational Cultural Competence Task Force. This task force, which will be chaired by Board member, **Tene Hamilton Franklin**, has been charged with developing a vision statement, creating and administering a climate survey, creating a strategic plan for continued education in cultural competence for all involved with NSGC and developing an evaluation strategy. The task force will conduct its work this summer, and the results will be presented at the NSGC Annual Education Conference in October.

Please look for and plan to complete the climate survey in August. If you have any questions or suggestions for the Board or the task force, please contact me (atrepani@wayne.edu) or Tene Hamilton Franklin (teneh@aol.com). We value your feedback as we move forward with this very important initiative.

Professional Development

This article is the second in a two-part series aimed at helping genetic counselors who have an abstract accepted for presentation at a conference. The first article, on designing

a successful poster presentation, appeared in the Spring issue of Perspectives. *This article details how to create and deliver an outstanding oral presentation. Both articles will be available to download from the Abstracts page on the NSGC website.*

Tips for Creating and Delivering a Great Oral Presentation

By Jehannine Austin PhD, CGC and Courtney Sebold MS, CGC

You have submitted an abstract to the conference you want to attend, and you have been selected to give a platform presentation. Great job! Now, you need to put your talk together.

Platform presentations are core components of most scientific meetings. They provide desired information to the audience, as well as the opportunity for attendees to showcase their work and exchange ideas with other scientists. It is important that oral presentations stimulate the audience and invite discussion. The goal of this article is to give genetic counselors some tips on creating an oral presentation that is informative, engaging and professional. Please note that these guidelines are not specific to the development of oral presentations for the NSGC Annual Education Conference, rather, these tips can be applied to the development of oral presentations in general.

What technology do I need in order to put together visual aids for an oral presentation?

Although it is possible to deliver a great oral presentation simply by using a chalkboard, most conferences will expect you to provide a computer-based visual aid for your presentation. There are many different slide presentation software programs available (e.g. Keynote for Macintosh), but Microsoft PowerPoint is the most commonly used program and is available for use with a PC or a Mac. The recommendations outlined in this article are the same regardless of what program is used to design the presentation.

What sort of content should I include in my presentation?

- Most importantly, KNOW YOUR AUDIENCE and design the content of your presentation accordingly. Do not explain inheritance patterns to a group of genetic counselors. However, if you are speaking to a group of family practitioners, it may be useful to briefly review basic genetic concepts.

- If you have an audience with varying levels of expertise, it is best to spend the majority of the time talking to the complete non-expert. For example, for a 10-minute presentation, spend only two minutes talking for those with more specialized knowledge.

- In order to make the material as accessible as possible, avoid inundating your audience with details and jargon.

- It is helpful to have an "Outline" slide for your talk. This shows the audience what you will be covering in your presentation. Similarly, it is always a good idea to conclude with a "Summary" slide. This slide reiterates the take-home messages of the presentation. This strategy can be summarized as:

    - tell them what you are going to tell them
    - tell them
    - tell them what you told them.

- If you are presenting research, be sure to acknowledge the individuals and funding agencies that were involved with the study. This slide may be placed at the beginning or end of the presentation.

- Presentations can be more engaging if you include some non-text slides between your scientific content. However, if you use picture or cartoon slides, make sure they are relevant to the subject matter. Interspersing your presentation with pictorial content that is not immediately relevant confuses your audience and interrupts the flow of the presentation.

How do I design my slides most effectively?

- To be effective, your presentation slides must be clear and easy to read by the entire audience. Keep in mind that you may be in a large conference room. For the main text of your slides, font size should be about 24 pt or larger. For titles, use a font size of 40-50 pt.

- Use standard horizontal formatting to avoid inadequate projection.

- Text slides should have single-spaced text, with a maximum of five lines. There should be no more than seven to eight words per line. Slides containing more words per line cannot readily be seen by the audience.

- Try to limit the amount of text on each slide by using bullet points.

- Use high contrasting colors for the background and text of color slides. For example, a blue background with white or yellow lettering contrasts nicely, whereas a blue background with red lettering is difficult to read.

- Use a font that does not have serifs (short lines that extend from the bottom or top of a letter). Fonts like Arial and Verdana have no serifs and are good choices for slides.

- Each slide should have a readily-identifiable main idea with limited additional information. Do <u>not</u> use complete sentences!

- Emphasize key points by using boldface, italics or different colors instead of underlining.

- If you have a slide with a table, try to limit the table to a maximum of three or four columns and five or six rows.

- If you have a slide with graphs, label the axes with large lettering, and highlight different curves with contrasting colors. Avoid more than two graphs on the same slide.

How many slides should I include in a presentation?

The amount of time you are given for your presentation will vary according to the conference. It is very important to adhere to this schedule. Exceeding the limit takes away from other presenters and does not allow for questions from the audience. In fact, at some conferences, moderators of oral sessions are instructed to cut presenters off if they go over their allotted time**.** So, remain professional and considerate, and be sure to adhere to your time slot.

- In general, you want to aim for one "content" slide per minute of presentation. For a 10-minute presentation, you would aim for a maximum of 10 "content" slides. Thus, the presentation would have a total of 13 slides, including the title, acknowledgements and outline slides.

- If you have too many slides and you have already limited the amount of text on each slide, you may be trying to present too much data. In this case, you need to consider the most important take-home message for your audience, and cut some material out of your presentation.

- If one of your "content" slides includes a table or graph, you may need to devote more than one minute to that slide to properly explain the graphic. Keep this in mind when planning your presentation.

- If you refer to the same slide more than once, use a duplicate slide. Do not ask the audience to recall a specific slide from earlier in the presentation. Do not flip back to a previous slide, as this can be difficult if you are nervous, and it is confusing for the audience.

How can I visually and vocally prepare myself to enhance my presentation?

- Practice and time your presentation beforehand to ensure that you will complete it within the allotted time.

- Try NOT to read your text from your slides. Instead, talk about the material that is on the slides.

- Speak clearly and slowly. Public speaking can cause people to talk too quickly if they are nervous or uncomfortable. Take a couple of deep slow breaths as you begin, and if you find yourself speeding up, pause briefly to take a deep breath or a sip of water.

- Be enthusiastic about the material you are presenting. A monotone voice can be very soporific.

- Engage your audience by making brief eye contact with various people around the room. Talk while looking out at the room rather than towards the floor or your notes.

- While your clothes are not the most important aspect of your talk, be sure to dress in a professional manner, appropriate for the style of meeting you are attending. Your put-together appearance can add to your validity as an expert on your topic.

Any tips for a nervous or first-time presenter?

- Practice the first few sentences/slides of your talk so that you know them by heart. You are often the most nervous when you first stand up, so having the first couple of slides perfectly planned can help you relax into the talk.

- Put a "friendly face" (a colleague or friend) somewhere in the audience so that you can readily see them from the podium. Look to them for encouragement and confidence during your presentation.

- Remember, you know more about the material you are presenting than anyone else in the room!

Summary

An informative and engaging presentation reflects the professionalism of the author and encourages discussions and collaborations with other scientists. For additional tips and advice on creating and delivering platform presentations, visit the NIH Virtual Career Center at: http://www.training.nih.gov/careers/careercenter/publish.html.

References:
Goldbort R. Professional Scientific Presentations. *Journal of Environmental Health*. 64(8):29-31. 2002.

For Your Practice Special Series: Cases in Expanded Metabolic Screening

This is the final article in a six-part series presented by the Metabolism/Lysosomal Storage Disease SIG in response to the expanded newborn screening panel developed in 2005 by the American College of Medical Genetics' Newborn Screening Expert Group[1]*. This panel comprises 29 conditions tested by all state screening programs, requiring genetic counselors to determine the impact and recurrence risks of unfamiliar metabolic conditions.* Perspectives *has highlighted several lesser-known genetic conditions now included in newborn screening to help genetic counselors as they come face-to-face with these diseases.*

Case 6: Carnitine Palmitoyltransferase 1A (CPT1A) Deficiency

By David M. Koeller, MD and Kelly Jo Hamman, MS

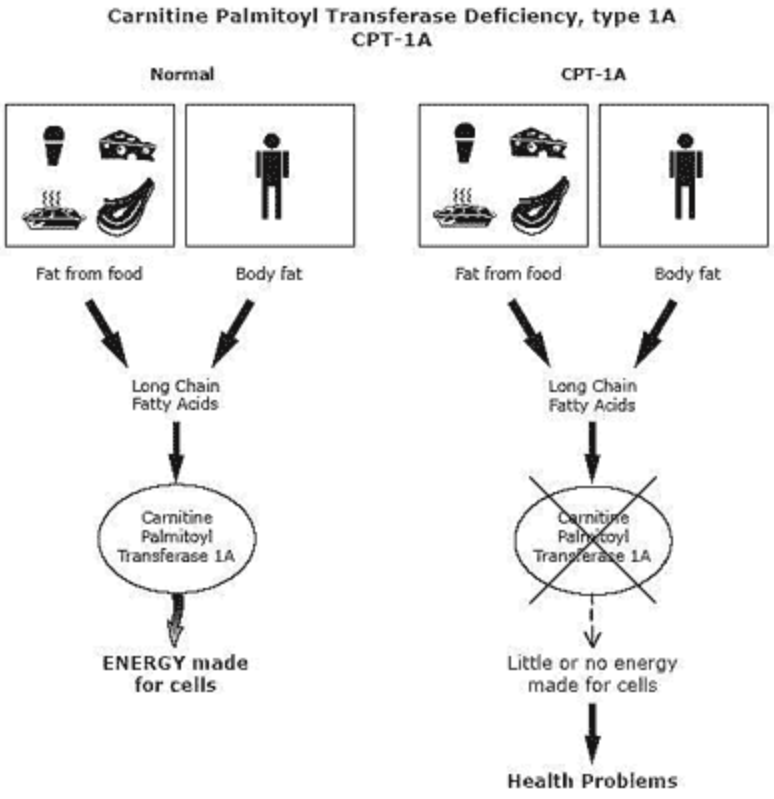
Disease Review

Biochemistry: Deficiency of Carnitine Palmitoyltransferase 1A (CPT1A), an enzyme that is required for the utilization of long chain fats as an energy source. In the liver, CPT1A is required for ketone production from long chain fatty acids during fasting. CPT1A deficiency is classified as a disorder of fatty acid oxidation.

Genetics: Autosomal recessive. The CPT1A gene is located at 11q13.

Incidence: Less than 1 in 100,000 in the general population. A common sequence variant (P479L) is found in ~20% of Alaska Natives and is also highly prevalent in the First Nations population of Canada and the Inuit populations of both Canada and Greenland.

[1] [1]Watson, MS, et al., "Newborn Screening: Toward a Uniform Screening Panel and System- Executive Summary", Pediatrics, Vol. 117 No. 5 May 2006, pp. S296-S307



Natural History: In the liver, CPT1A is required to metabolize long chain fats and generate ketones. Patients with complete CPT1A deficiency run out of fuel (glucose and ketones) during a prolonged fast, resulting in metabolic decompensation. Affected patients typically present within the first three years of life with symptoms that include hypoketotic hypoglycemia, hepatomegally and elevated liver enzymes. Hyperammonemia, seizures and coma can also be seen. As with other fatty acid oxidation disorders, it can also result in sudden unexplained infant death (SIDS). In infants and young children symptoms can be precipitated by any illness, such as gastroenteritis, that results in fever and reduced oral intake. Recurrent or even isolated severe episodes of metabolic decompensation can result in permanent neurodevelopmental problems and seizures. Fortunately, symptoms can be prevented by the avoidance of fasting and the institution of appropriate therapy, including intravenous glucose, during illness and other times of high risk. The ability to avoid symptoms by appropriate therapy in patients known to have CPT1A deficiency underlies the importance of including this disorder in expanded newborn screening panels.

The form of CPT1A deficiency common in the Alaska Native population is due to a P479L sequence variant in the CPT1A protein sequence, which results in only a partial loss of enzyme activity. Preliminary evidence suggests that most affected infants and children never have any symptoms. However, some patients have presented with severe symptoms as described above. Currently no unique risk factors have been identified in the Alaska Native population that allow for prediction of who is most likely to develop symptoms, and therefore, all families are counseled about the risks of fasting and how to avoid and/or treat symptoms.

Genetic Counseling - Challenges and Solutions in the Alaskan Native Population

CPT1A deficiency was first identified in the Alaska Native population in early 2004. At that time confirmatory testing to follow up a positive newborn screen required a skin biopsy to establish a fibroblast culture, which was used to measure the enzymatic activity of CPT1A. The remote location of many of the affected infants, who often live in villages that are served only by a health aid that visits a few times a year, made this extremely difficult. Fortunately, the determination that all of the affected infants were homozygous for the same sequence variant (c.1436C→T) allowed DNA testing to be used for confirmation of an abnormal newborn screen.

As the numbers of affected infants continued to grow, the ability to evaluate all of the infants and counsel their families became an issue. There is no metabolic clinic based in Alaska, and the outreach clinics staffed by providers from the University of Washington could not handle the volume of patients being identified. Therefore, in 2007 Dr. David Koeller from the Oregon Health & Sciences University started a CPT1A clinic at the Alaska Native Medical Center in Anchorage. However, in the fall of 2007 it was determined that the incidence of CPT1A deficiency in Alaska Native infants was far higher than what was being observed by expanded newborn screening and could possibly be as high as 600 affected infants per year, which would overwhelm the existing CPT1A clinic. This realization is forcing the development of new approaches to patient management and family counseling. The method used for newborn screening for CPT1A deficiency also is being re-evaluated.

One of the greatest challenges that has arisen since learning of the high prevalence of CPT1A deficiency due to the c.1436C→T sequence variant in the Alaska Native population is determining whether it truly represents a disease or merely a benign variant. It is clear that most affected infants, children and adults never have symptoms. Additional unidentified risk factors, both genetic and environmental, are likely important.

Teaching Lessons

1) Accurate knowledge of natural history is essential for effective newborn screening.

Expanded newborn screening has the potential to identify disorders about which we have inadequate knowledge to accurately counsel families on management and long-term outcome. We must strive to avoid adding the anxiety resulting from such uncertainty to the list of concerns facing new parents, who may already feel overwhelmed with their new baby. In the case of CPT1A deficiency in the Alaska Native population, this uncertainty affects not just individual parents but entire Native communities whose members are affected. It also is a significant concern for the Alaska Department of Health, which is responsible for assuring that all babies born in Alaska receive the care they require. Determination of the true health consequence of CPT1A deficiency in the Alaska Native population is an urgent priority.

2) Cultural sensitivity must be incorporated in all aspects of care.

The identification of a disease or even a DNA sequence variant within a specific population has the potential to result in the labeling of members of that group as being somehow inferior or weak. Awareness of the potential for stigmatization is necessary for the development of effective, culturally sensitive approaches for care of affected individuals. When working with a population such as Alaska Natives, members of the affected community should be involved in all aspects of program development and to seek formal approval before implementation.

Legislation Report

Are You Working on Licensure? Let NSGC Help!

Have you started to talk about licensure in your state? Not sure how or where to start the process? Do you have a bill ready to be introduced? NSGC wants to help. We can review your bill language and advise on issues that arise in the licensure process. It is important that NSGC be involved in this process, especially in the early stages for several reasons:

- It will ensure the language in your bill is consistent across states to allow for reciprocity.
- It will be necessary for NSGC to write a letter of support on behalf of licensure in your state.
- NSGC can be a resource in navigating the legislative process. Since the legislative process often moves very rapidly, NSGC's prior knowledge of your bill's language will allow us to respond quickly to legislators' requests and to expedite our assistance in issues or requests that you may have.

Let us help you achieve licensure. Contact the Licensure Subcommittee, Cheryl Harper at CHARPER@beaumonthospitals.com, John Richardson at jrichardson@smithbucklin.com, or Amy Crunk at amy624@gmail.com, to let us know where you are in the process of state licensure and if you have any questions that we can help answer.

"Having NSGC review our bill language before it was introduced enabled our legislative process to go smoothly. They made suggestions to make our bill stronger and helped us deal with the issue of being regulated by the Board of Medical Examiners instead of being able to create our own board. The Licensure Grant we received was a big help in paying for our website and the copying of materials for legislators."

---Martha Dudek, Tennessee

"I want the membership to know that John Richardson and Cheryl have been consistently accessible and a tremendous source of knowledge and support as we navigate through the licensure hurdles in New Jersey. John, at a moment's notice, has provided feedback, re-written portions of our bill, as well as letters/memos to legislators and coached me prior to any testimony that I had to provide both in the New Jersey Senate and House. Whether we faced the hurdles of opposition from pro-life groups, financial opposition, issues of registration vs. licensure, etc., etc., John has provided me with the knowledge and the language necessary to articulate and thus advocate effectively on behalf of licensure. I encourage all GCs involved in licensure in their state to utilize the valuable resources that NSGC offers."

---Elena Ashkinadze, New Jersey

NSGC News

Governance Evaluation Task Force

A Task Force has been convened to evaluate the governance changes approved by the NSGC Board of Directors in 2007. It consists of members of the original Task Force to ensure memory of the original intent of the changes, members who were added during the implementation phase, and new individuals to add a fresh and objective perspective. The Governance Evaluation Task Force members include:

Cathy Wicklund, Chair
Luba Djurdjinovic
Tene Hamilton Franklin
Steve Keiles
Peter Levonian
Robert Pilarski
Angie Trepanier
Vickie Venne
To view the members of the original task force and the implementation task force, please click here.

Evaluation of NSGC's governance changes and the success with moving from the previous to the new governance structure will be ongoing. Preliminary evaluation will focus on whether full implementation of the changes has been achieved and evaluating member participation in the new structure. In this initial phase, the process will concentrate on the areas most affected by the governance changes. In the future, evaluation will be expanded to include ongoing monitoring of NSGC's overall governance and recommendations for refinement of any changes to ensure that they are working as intended. The primary areas of evaluation for 2008 are:

- Committee Evaluation
- Nominations Process Evaluation
- Liaison Evaluation
- SIG Evaluation
- NSGC Board of Directors and Society as a Whole

The Task Force has identified particular items of consideration for each area of evaluation, as well as metrics for evaluating. Monitoring for each of these areas has been ongoing.

Most recently, the Task Force met to assess the progress of Committees toward fulfilling the charges for 2008; feedback from the Committee Chairs and Vice Chairs about the new structure; and communication between the Board and Committees through the Board Liaisons. NSGC's Committees are making great progress towards their annual charges and tasks. Additionally, the Task Force has monitored how the Committees are using the Leader/Volunteer database to engage additional members. While use of the Leadership/Volunteer Database varies from Committee to Committee, all committees have used the database at least once to tap into additional volunteers. If you are interested in volunteering for NSGC but have not yet submitted a Willingness to Serve form, we encourage you to do so as this is a great way to let Committee Chairs and others know that you are interested in being contacted as projects arise. The form is available on NSGC's Leadership/Volunteer Development web page at http://www.nsgc.org/members_only/leader_volunteer_program/index.cfm

More comprehensive evaluation of the success of the governance changes will be performed after a full year has passed. True evaluation must take place over several years to assess the full effect of the changes. As always, if you have comments regarding NSGC's governance changes or current governance structure, please feel free to contact any member of the Governance Evaluation Task Force. We welcome your feedback!

SIG Speak

GINA - 13 Years in the Making

By Elizabeth Hoodfar, MSc, (C)CGC, Jennifer Leib, MS, CGC and Co-Chairs Rebecca Nagy, MS, CGC and Joy Larsen Haidle, MS, CGC, on behalf of the Cancer SIG of NSGC

Thirteen years ago, Representative Louise Slaughter (D-NY) and Senator Olympia Snowe (R-ME) introduced the Genetic Information Nondiscrimination Act (GINA) to the 104th Congress (1995-96). Since then various iterations of legislation to protect individuals against genetic discrimination by insurers and employers have been introduced and debated in the House of Representatives and/or in the Senate. Remarkably, GINA had not passed both Houses until April 2008 (the 110th Congress). In May it was signed into law by the President.

Patient Protections

The protections of GINA are afforded to any individual seeking presymptomatic genetic information. Like all of NSGC, the Cancer SIG received the Public Policy Committee's call of action to support GINA efforts in the last week of Senate deliberations, and we thank all those who responded. We also received a number of stories from patients who had concerns about genetic discrimination. Aspects from several of these cases were used during **Congresswoman Biggerts'** floor statement prior to the House of Representatives' vote. One woman's story about hereditary cancer susceptibility highlights how concerns of genetic discrimination affected the decisions she and her family made. In her own words:

"After my second cancer diagnosis (colon at age 31, uterine at age 48), and on the advice of my oncologist, I decided to have a blood test to see if there was a genetic cause. The test came back positive, a gene mutation was found. I'm not really any different from most of the other people walking around on this planet, because almost everyone carries a risk or genetic pre-disposition to some kind of health problem or disease. My situation is unique because I know about it. I am aware of my genetic predisposition to cancer and can work with my doctors to plan my preventive healthcare accordingly.

I consider this to be a huge advantage over the average person who has no idea what health issues could be heading their way. I think of my aunts, uncles, grandparents, parents - some of them have also had this gene but their lives were cut short because they didn't know. What a shame that I have to be concerned about with whom I share the cause of my illnesses. This is information that I am blessed to have and should be able to use to my benefit, not my detriment. I'm lucky to be in [a state] where I'm guaranteed healthcare coverage, but what if I move, leave or somehow my situation changes? What if I want to buy more life insurance? And what about my children that may have inherited this gene? So I am a closet mutant. I talk with my physicians about it, but we are keeping it out of my charts. It will stay that way for me and my kids until we know there won't be a penalty to pay for trying to survive."

GINA can go a long way to alleviate many of this woman's concerns.
Her genetic test result could not legally be used to discriminate against her or any of her family members in group or individual health insurance, or in employment practices/benefits. These minimum protections would follow her and her family members regardless of the state they lived or worked. GINA cannot, however, afford her or her relatives any protection against discriminatory practices in obtaining life insurance, disability insurance, long-term care or critical care insurance.

Legislation Then and Now

In 2004, the National Partnership for Women & Families, on behalf of the Coalition for Genetic Fairness, published a report titled, "Faces of Genetic Discrimination – How Genetic Discrimination affects Real People" (www.nationalpartnership.org). This document gave an overview of fears, costs and the impact of genetic discrimination, cited case examples and summarized the broad spectrum of support for anti-discrimination laws.

To date there are 41 states with genetic nondiscrimination laws for insurance and 32 states with such laws for employment. Federally there is a patchwork of protections from various sources, including the Americans with Disabilities Act (ADA,1990), the Health Information Portability and Accountability Act (HIPAA, 1996), the Executive Order (2000) issued by President Clinton and the Equal Employment Opportunity Commission (EEOC,1965). However, given the variability in approach and application of these laws, all are incomplete in the scope and depth of protection offered, individually and collectively.

Congress has declared that the intent of GINA is to provide "a national and uniform basic standard" for the full protection of the public from discrimination and thereby allow "individuals to take advantage of genetic testing, technologies, research and new therapies." **Francis Collins**, director of National Human Genome Research Institute (NHGRI), has called the passage of GINA "a great gift to all Americans." It is the hope of all genetic counselors that we can use GINA to relieve our patients of genetic discrimination fears and allow them to open the doors of opportunity that genetic information presents.

Summary of GINA

GINA has three main sections:

> Title I: Genetic Nondiscrimination in Health Insurance
> Title II: Prohibiting Employment Discrimination on the Basis of Genetic Information
> Title III: Miscellaneous Provisions (which is standard housekeeping language for any bill).

Key terms used within GINA are defined as follows:

Genetic information: An individual's or family member's genetic testing or the manifestation of a disease in family members. This includes any request for or receipt of genetic services or the participation in clinical research that involves genetic services.

Genetic test: Any analysis of human DNA, RNA, chromosomes, proteins or metabolites that detects genotypes, mutations or chromosomal changes.

Genetic services: A genetic test, genetic counseling (including obtaining, interpreting or assessing genetic information) or genetic education.

The terms "pedigree" and "family history" are not specifically used within GINA. It could be argued that these terms are incorporated either under the broad definition of "genetic information" or in the definition of "genetic counseling" under "genetic services."

Title I: Genetic Nondiscrimination in Health Insurance

This section prohibits group health plans, issuers of health insurance to individuals and issuers of Medicare supplemental policies from using genetic information to:

- adjust premium fees or contribution amounts
- establish eligibility rules for enrollment
- condition issuance or effectiveness of a policy
- impose a preexisting condition exclusion.

These entities also are prohibited from requesting, requiring, using, disclosing or purchasing genetic information concerning an individual or a family member for underwriting purposes. This prohibition applies to the use of genetic information about a fetus or embryo for underwriting related to the pregnant woman, the legal owner of embryos created by assisted reproductive technologies or their family members.

Title II: Prohibiting Employment Discrimination on the Basis of GeneticInformation

This Title applies to employers, employment agencies, labor organizations and joint labor-management committees. The provisions make it an unlawful employment practice to discriminate against an individual based on genetic information. Discrimination includes:

- failing to hire, discharging or failing or refusing to refer for employment
- treating differently with respect to compensation, terms, conditions or privileges of employment
- treating differently in admission to apprenticeships, training or retraining programs.

This Title also prohibits employers and other entities named above from obtaining genetic information (with certain exceptions, such as in forensic DNA labs for identifying sample contamination) and requires that genetic information be maintained as confidential in separate files. However, a law suit cannot be brought under GINA if someone suffers "disparate impact" (i.e. is adversely affected and suffers negative consequences) on the basis of genetic information.

Impact of GINA

GINA will be enacted once the Secretary of Labor issues final regulations on how to implement and apply the law. The provisions of Title I will be effective one year after GINA's enactment. The provisions of Title II will be effective 18 months after enactment.

Only time will tell how much and to what extent the benefits of GINA will be realized for the public, as well as for the medical and scientific community. Thank you to all who have worked tirelessly over the past 13 years to pass this significant legislation.

For more information on GINA, go to the National Human Genome Research Institute at http://www.genome.gov/PolicyEthics/.

A full text of GINA (H.R. 493) is available at http://thomas.loc.gov/cgi-bin/bdquery/z?d110:HR00493.

Genetic Counselor Publications

By Deborah McDermot, MS, CGC and Alisha Biser, MS, Sarah Lawrence College 2nd year student

Featured Papers:

Austin JC, Honer WG. Psychiatric genetic counseling for parents of individuals with psychotic disorders: A pilot study. *Early Intervention in Psychiatry*. 2:80-89. 2008.

Peay HL, Palmer C, Sheidley B, McCarthy Veach P, Gettig B, Austin JC. Psychiatric disorders in clinical genetics I: Assessing family histories of psychiatric disorders. *J Gen Couns*. 17(1):6-17. 2008.*

Austin JC, Palmer C, Sheidley B, McCarthy Veach P, Gettig B, Peay HL. Psychiatric disorders in clinical genetics II: Individualizing recurrence risks for psychiatric disorders. *J Gen Couns*. 17(1):18-29. 2008.*

Psychiatric conditions are common in the general population, and it is not unusual for most genetic counselors to encounter clients with personal or family histories of psychiatric illness. The extent to which genetic counselors should address these family histories with their clients remains uncertain, however, as there is a paucity of information on this subject in the genetic counseling literature.

After examining the issues brought forth from the 2006 NSGC Educational Breakout Session presented by the Psychiatric Special Interest Group (SIG), **Jehannine Austin** and her colleagues, including SIG Co-Chair and co-author, **Holly Peay**, recently created a two-part professional development series addressing these particular issues. The first article explains the generation of recurrence risks based on empiric risk data. The second article discusses the generation of individualized recurrence risks and methods of managing the uncertainties of affected individuals and families. Together these articles provide a framework for genetic counselors who so often are challenged by the discussion of mental illness with patients. Jehannine draws on her own personal experience and expertise in the area of psychiatric counseling, adding valuable

recommendations to readers.

Jehannine began her career in neuropsychiatric genetics as a PhD candidate, and throughout the course of her training, she met with families affected with psychiatric disorders. While equipped with the tools to discuss rudimentary details of what contributes to the development of psychiatric illness, she felt she did not have the language to adequately discuss the varying conditions in terms her patients could understand. Consequently, after the completion of her PhD, she entered a genetic counseling training program to gain this expertise. Since her graduation in 2003, she has continued to work exclusively with psychiatric genetics. The article she published on a pilot study of counseling for parents of those affected with psychotic disorders illustrates the components of her training coming together.

Jehannine is currently an Assistant Professor in the Department of Psychiatry at the University of British Columbia (UBC). Her position is funded by national support from the Canadian Institutes of Health Research and a provincial grant from the Michael Smith Foundation for Health Research Institute. She educates the UBC genetic counseling trainees, medical students and residents. Additionally, she increases public awareness of psychiatric conditions by speaking at local organizations like the BC Mood Disorders Association and the BC Schizophrenia Society. Through her community work, she has noticed a striking discrepancy; while many people with psychiatric illnesses and their family members have indicated that they would like genetic counseling, only a small proportion have actually received it. Her current research, along with genetic counselor and fellow SIG member, **Catriona Hippman**, focuses on the response to her novel method of actively offering genetic counseling services in the community.

In her articles, Jehannine discusses how genetic counselors are suited for providing a supportive forum in which to explore the impact of psychiatric illness. Considering that the genetics of psychiatric conditions are not fully understood, difficulty arises in interpreting individual risks. However, psychiatric genetics, she believes, is a fast-paced arena for continuing development. For example, she anticipates that psychopharmacogenomics will eventually be achieved, better individualizing treatment options, which may decrease, if not eliminate, the burdensome side effects of current therapies.

When more is discovered about the genetic etiology of psychiatric conditions, genetic counselors will play a critical role in helping patients understand these conditions and interpret risk assessments. Jehannine encourages genetic counselors to explore this untapped area; she believes that increased emphasis on psychiatric disorders in genetic counseling curricula will enhance recruitment into this specialty.

**Note: JGC articles are not typically included in these publications listing. In this issue, they were highlighted contributing to the interview with Jehannine Austin.*

Articles Co-Authored by Genetic Counselors Dec 2007-May 2008

(names of genetic counselors appear in bold)

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

Bradley LA, **Kloza EM**, Haddow PK, Beauregard LJ, Johnson JL, Haddow JE. A genetic history questionnaire-based system in primary prenatal care to screen for selected fetal disorders. *Gen Testing*. 11(3):291-295. 2007.

Brooks BP, MacDonald IM, Tumminia SJ, Smaoui N, **Blain D**, Nezhuvingal AA, Sieving PA. Genomics in the era of molecular ophthalmology: Reflections on the National Ophthalmic Disease Genotyping Network (eyeGENE). *Arch Ophthalmol*. 126:424-425. 2008.

Duncan RD, **Prucka S**, Wiatrak BJ, Smith RJ, Robin NH. Pediatric otolaryngologists' use of genetic testing. *Arch Otolaryngol Head Neck Surg*. 133(3): 231-236. 2007.

Faucett WA, Hart S, Pagon RA, Neall LF, Spinella G. A model program to increase translation of rare disease genetic tests: Collaboration, education and test translation program. *Gen Med*. 10(5): 343-348. 2008.

Harris J, **Fraser J**. "Genetics of Movement Disorders, Chpt. 32." *The Molecular and Genetic Basis of Neurologic and Psychiatric Disease, 4th ed*. Rosenberg R et eds. 2007.

Howard KL, Hall DA, Moon M, Agarwal P, Newman E, Brenner M. Adult-onset Alexander disease with progressive ataxia and palatal tremor. *Mov Disord*. 23(1):118-22. 2008.

Hwang ES, McLennan J, Moore DH, **Crawford BB**, Esserman LE, Ziegler JL. Ductal carcinoma in situ (DCIS) in BRCA mutation carriers. *J Clin Oncol*. 25:642-7. 2007.

Kaplan KD, Brown M, Croughan MS, Turek PJ. The relative accuracy of a questionnaire compared with pedigree analysis in genetic risk assessment of infertility. *J Urol*. 179(4):1499-505. 2008.

Ledbetter DH, **Faucett WA**. Issues in genetic testing for ultra-rare diseases: Background and introduction. *Genet Med*. 10(5):309-313. 2008.

Labay V, Garrido G, **Madeo AC**, Nance WE, Friedman TB, Friedman PL, Del Castillo I, Griffith AJ. Haplogroup analysis supports a pathogenic role for the 7510T>C mutation of mitochondrial tRNA (Ser(UCN)) in sensorineural hearing loss. *Clin Gen*. 73(1):50-4. 2008.

Lubitz-Gordon R, Komaromy M, **Crawford B**, **Lee R**, Luce J, Ziegler J. An evaluation

of novel genetic educational materials for an ethnically diverse population. *Gen Testing*. 11:276-290. 2007.

Mikhail FM, Sathienkijkanchai A, Robin NH, **Prucka S**, Biggerstaff JS, Komorowski J, Andersson R, Bruder CE, Piotrowski A, de Stahl TD, Dumanski JP, Carroll AJ. Overlapping phenotype of Wolf-Hirschhorn and Beckwith-Wiedemann syndromes in a girl with der(4)t(4;11)(pter;pter). *Am J Med Gen A*. 143(15):1760-1766. 2007.

Mitchell BD, McArdle PF, Shen H, Rampersaud E, **Pollin TI**, Bielak LF, Jaquish C, Douglas JA, Roy-Gagnon MH, Sack P, Naglieri R, Hines S, Horenstein RB, Chang YP, Post W, Ryan KA, Brereton NH, Pakyz RE, Sorkin J, Damcott CM, O'Connell JR, Mangano C, Corretti M, Vogel R, Herzog W, Weir MR, Peyser PA, Shuldiner AR. The genetic response to short-term interventions affecting cardiovascular function: Rationale and design of the Heredity and Phenotype Intervention (HAPI) Heart Study. *Am Heart J*. 155(5):823-8. 2008.

McInerney JD, **Faucett WA**. PAs and the practice of genetic medicine. *JAAPA*. 21(5). 2008.

Morales A, **Cowan J**, Dagua J, et al. The family history: An essential tool for cardiovascular genetic medicine. *Congest Heart Fail*. 14:37-45. 2008.

Morales A, Dagua J, et al. Genetic testing and genetic counseling in cardiovascular genetic medicine: Overview and preliminary recommendations. *Congest Heart Fail*. 14(2):97-105. 2008.

Nauman D, **Morales A**, **Cowan J**, et al. The family history as a tool to identify patients at risk for dilated cardiomyopathy. *Prog Cardiovasc Nurs*. 23(1):41-44. 2008.

Prucka SK, **McIlvried DE**, Korf BR. Cancer risk assessment and the genetic counseling Process: Using hereditary breast and ovarian cancer as an example. *Med Princ Pract*. 17:173-189. 2008.

Peters LM, Fridell RA, Boger ET, San Agustin TB, **Madeo AC**, Griffith AJ, Friedman TB, Morell RJ. A locus for autosomal dominant progressive non-syndromic hearing loss, DFNA27, is on chromosome 4q12-13.1. *Clin Genet*. 73(4):367-72. 2008.

Sathienkijkanchai A, **Prucka SK**, Grant JH, Robin NH. Isolated facial hemihyperplasia: manifestation of Beckwith-Wiedemann syndrome. *J Craniofac Surg*. 19(1): 279-83. 2008.

Uhlmann WR, Guttmacher AE. Key internet genetics resources for the clinician. *JAMA*. 299:1356-1358. 2008. *(this was a special theme issue of JAMA focused on genetics and genomics)*

Velinov M, **Zellers N**. The fetal mycophenolate mofetil syndrome. *Clin Dysmorphol*. 17(1)77-8. 2008.

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

AEC Update

Something for Everyone!

Check your mailboxes soon for your official conference booklet detailing the 27th NSGC Annual Education Conference (AEC) and 2008 Short Course in Los Angeles, CA. The NSGC membership has grown significantly in the number of members and the variety of professional environments in which we practice. As such, the AEC will feature a wide variety of content focusing specifically on the genetic counselor in the hopes of addressing the educational and professional needs of our diverse group.

Taking Heredity to Heart

The 2008 Short Course "Taking Heredity to Heart: Cardiovascular Genetics, An Overview," was chosen for its timeliness and relevance to our professional and personal lives. Cardiovascular diseases mark the leading causes of death in the United States and are a growing public health concern worldwide. Our understanding of the genetic causes and risk factors for cardiovascular disease is increasing rapidly, and the demand for genetic counseling services for this class of diseases is expanding. Attendees will gain significant knowledge in areas such as the impact of genetics on the practice of cardiology, familial evaluation of atherosclerosis, incorporating cardiogenetics into your practice and the multiple facets of managing a cardiogenetics family.

Outreach in LA

In an effort to reach out to the community of our local host city, the NSGC annually conducts an Outreach Event during the AEC. This year's event will involve local college and nursing students networking with genetic counselors from a variety of settings in a fun and informal atmosphere. Please consider participating in this stimulating event – stay tuned to www.nsgc.org for more details as they become available.

Prepare for Fun!

The 2008 AEC and Short Course are being held at the Hyatt Regency Century Plaza – a prime location approximately 10 miles from Los Angeles International Airport and across the street from the Westfield Shopping Center with over 18 stores, a movie theater, six

full service sit-down restaurants and a "fine dining" food court. In January 2007, the Hyatt completed a $40 million renovation including all guest rooms and meeting spaces, an upscale Equinox Fitness Center, a full-service Starbucks and the X-Bar, an indoor/outdoor lounge open daily from 4:00 pm to 2:00 am. Discounted room rates are subject to availability, so be sure to book your room today.

For more information on exciting things to do while you are in Los Angeles, please visit the AEC web page on www.nsgc.org.

Many Thanks

We would like to thank our conference committee co-chairs – Jehannine Austin, Courtney Sebold, Lynn Holt, Brooke Smith, Beth Wood, Christine Stanislaw, Kimberly Wendt and Amy Crunk – as well as their committee members, to whom we owe a huge debt of gratitude. This conference is the result of the tireless efforts of this outstanding committee. Without each and every one of these individuals, this conference would not be possible. We look forward to seeing you in Los Angeles!

Janice Berliner, JBerliner@sbhcs.com
Stephanie Brewster, stephanie.brewster@childrens.harvard.edu
2008 NSGC AEC Co-Chairs

Student Forum

BU Forges a Successful Path in Its First Few Years

By Lindsay Paull, BA, MS

The year 2008 has brought many new and exciting accomplishments for the Boston University School of Medicine Genetic Counseling program. In February, BU's director, **MaryAnn Whalen Campion**, received word from ABGC that the program was granted Full Accreditation. I am pleased recognize to the many innovative and intellectual minds that have collaborated to make this accomplishment possible. Congratulations to all of the local genetic counselors and the Boston University faculty who have defined the program.

Theses Accomplished

In mid May, BU graduated its second class of students. This small class of five was thrilled to present their thesis projects at the end of April. Each student focused on their own area of interest with their research topics including:

Quality of Life Issues with Costello Syndrome
Patient Perceptions about Prenatal Genetic Diagnosis

Issues Surrounding Genetic Counseling and Adoption
Educational Approaches for Children with Gaucher Disease.

In addition, one student created a highly successful educational tool for mitochondrial disorders and is hopeful to publish and distribute her aids to the genetic counseling and mitochondrial community in the near future.

Commitment to Community

These thesis projects are just one of the many ways that BU's students have become immersed in the genetic counseling community. Over the past two years, the students have participated in several community and advocacy related events. They have educated families at the Perkins School for the Blind, participated in the annual Miles for Mito 5K run and visited day care programs and community homes for individuals with disabilities. The students also invested tremendous effort in their rotations, ranging from local Boston-based hospitals to those in central Massachusetts, Rhode Island and southern New Hampshire. The counselors and geneticists who supervised at these hospitals have been integral to the students' academic and professional development and are greatly appreciated.

As the second-year class prepares for life beyond graduate school, the first-year class is ready to step into a senior role. Similar to their successors, these students embraced community involvement, volunteering at the Huntington's disease walk this past fall, participating in the Boston Medical Center Halloween Town to benefit the Kid's Fund, contributing to the Harvard All University Relay for Life in April, teaching a class about DNA to the Science Club for Girls in Newton, MA and attending various support group meetings around the area. They are now preparing for their upcoming summer internship rotations in locations that span from New England to Alaska.

A New Generation

The previous classes of BU students have paved the way for a growing and thriving genetic counseling program. Seven new students have been accepted to begin in the fall, traveling to Boston from hometowns in India, Oregon and Massachusetts. As this fourth class arrives, the faculty continues to strengthen the program by incorporating the innovative ideas of the former students and anticipating the fresh perspectives of new students.

Book Review

NEEDS PHOTO OF BOOK COVER

The Power of Two: A Twin Triumph over Cystic Fibrosis

Authors: Isabel Stenzel Byrnes and Anabel Stenzel

Publisher: University of Missouri Press, Columbia, MO, 2007*Reviewed by:* Bryanna Cox, MS, CGC

The Power of Two: A Twin Triumph over Cystic Fibrosis chronicles the life of identical twin sisters, Isa and Ana, who have cystic fibrosis. Their memoir is written by both women, who alternate chapters to tell how they live a full life in spite of having a chronic, limiting genetic disease.

Challenges of Chronic Illness

Ana and Isa were diagnosed with cystic fibrosis shortly after their birth. The women candidly write about the challenges their parents faced raising two ill children and the toll this had on the family. Their vivid descriptions of the time-consuming and physically exhausting daily treatments illustrate how difficult it can be to cope with chronic illness.

As young adults, both women developed advanced lung disease and underwent lung transplantation. Ana's recollection of Isa's critical condition immediately prior to her lung transplant is especially powerful. Both women share their experiences of meeting with the families of their donors and share the complex emotions involved with getting the chance to live due to another's death.

A Special Bond

As identical twins, the girls shared a special bond. But their bond was made stronger by their illness, as the girls relied on each other for support, encouragement and health care needs. As they became older, they performed their daily treatments on each other to relieve this burden from their parents. They attended the same university and were roommates, so they were available to help each other. At times, their relationship was strained as they struggled for personal identity in spite of their dependence upon one another. As the girls matured, their relationship evolved as they developed individual careers and outside intimate relationships.

For A Broad Audience

This book can be valuable resource to any individual with a serious disease who is struggling to find hope and maintain a commitment to living a full life. The candid way the women describe their emotions and experiences regarding cystic fibrosis will likely resonate with patients and families affected by many genetic conditions. This book also can provide genetic counselors better insight into the challenges and joys these families encounter.

Note: Isabel Stenzel Byrnes and Anabel Stenzel will discuss this book and their experiences with cystic fibrosis at the 28th Annual Education Conference this fall in Los Angeles. See your program book for details.

Research Network

The PTEN Study

The PTEN Study at the Cleveland Clinic is actively recruiting persons with confirmed or suspected diagnoses of Cowden syndrome, Bannayan-Riley-Ruvalcaba syndrome, Proteus syndrome or Proteus-like syndrome for research testing. The purpose of this study is to determine the minimal number of features associated with PTEN alterations at the DNA, RNA and/or protein levels. Patients who have had a positive PTEN result through a clinical laboratory also are welcome to join.

At the DNA level, our study first analyzes the PTEN gene through DGGE and sequencing of the PTEN promoter and coding sequence and adds MLPA of all PTEN exons for those patients meeting NCCN diagnostic criteria or with a BRRS phenotype. Clinically relevant results will be made available to the referring provider. Persons with a PTEN mutation, variant or polymorphism detected through our lab will be eligible for a subsequent questionnaire study. We hope that our research will contribute to the development of treatments and therapies for those with conditions in the PTEN spectrum.

Contact: http://www.lerner.ccf.org/gmi/research/pten/

Hereditary Basis of Neural Tube Defects

The Duke Center for Human Genetics is recruiting families in which one or more members have any type of neural tube defect, including spina bifida and anencephaly. Study participation includes a telephone interview, a review of medical records of the pregnancy and/or confirmation by family member(s) with an NTD of the type and level of the lesion, and collection of amniocytes, tissue, cord blood or blood samples from the pregnancy or person with the NTD and his or her parents and siblings, if possible. Chromosome analysis is performed on anencephaly cases only, and these results are shared with the family.

Contact: The Duke NTD Study, (866) 385-3683, ntd@chg.duhs.duke.edu, http://www.chg.duke.edu/diseases/ntd.html

Genetic Cardiology Research

Dr. Bruce Gelb, Director of The Center for Molecular Cardiology at Mount Sinai School of Medicine, is seeking individuals for ongoing gene identification projects related to congenital heart defects, Noonan syndrome and CHAR syndrome. Mount Sinai also is an approved site for the Marfan Trial, a study of Beta Blocker Therapy (Atenolol) versus

Angiotensin II Receptor Blocker Therapy (Losartan) in individuals with Marfan syndrome.

Contact: Kerri Lee, MS, (212) 241-6012, kerri.lee@mssm.edu

Fragile X Mutation Study

Patients and families with mutations in the *FMR1* gene are invited to participate in a collaborative project between the Institute for Basic Research (IBR) and Genzyme Genetics on intermediate and pre-mutation fragile X alleles.

The specific aims of the study are to conduct an epidemiological study of allele stability by examining repeat size distributions in families and to examine factors that may influence repeat instability. The index case must carry an *FMR1* allele with 45 to 200 repeats. One or both parent(s) and siblings also are asked to enroll. The referring provider will offer participation to patients. Blood draw kits and shipping supplies are provided at no cost; the family is responsible for blood draw fees. International participants may be considered on an individual basis.

Molecular testing is performed at IBR, and initial results may take one to two months. Results will be sent to the referring physician/genetic counselor. Other at-risk family members are then eligible for participation. Prenatal samples will not be accepted, but a sample from the baby may be submitted postnatally. Pregnant patients may enroll, but all prenatal testing must be handled separately.

Contact: Marcia Jodah, MS, CGC, (813) 250-0588, marcia.jodah@genzyme.com

Familial Pancreatic Cancer Genetic Study

The Karmanos Cancer Institute/Wayne State University is seeking volunteers for a genetic study of familial pancreatic cancer. Eligible families must have two or more individuals diagnosed with pancreatic adenocarcinoma. Affecteds persons need not be alive, however, DNA from at least one affected must be available via blood, buccal or archived tissue samples. Participants must be 18 years of age or older and must provide family/medical/lifestyle information and submit a blood sample. Travel is not required. Participants are not guaranteed to receive "gene test results," however, they can choose to be re-contacted should information arise from the study that may impact health behavior.

This study is conducted by the Genetic Epidemiology of Pancreatic Cancer (PACGENE) Consortium: Karmanos Cancer Institute/Wayne State University, Mayo Clinic, University of Texas/MD Anderson Cancer Center, Johns Hopkins University, University of Toronto/Mount Sinai Hospital and Dana-Farber Cancer Institute. Grant funding has been received from the National Cancer Institute.

Contact: Kate Sargent, MS, CGC, Family Project Coordinator, (313) 578-4240, sargentk@med.wayne.edu

Two Studies Investigating Dystonia

Volunteers are needed for dystonia research conducted at Massachusetts General Hospital.

1. Genetics of Dystonia

   Researchers are searching for new genes related to dystonia. Individuals diagnosed with any type of dystonia may participate including:

   - generalized/torsion
   - dopa-responsive
   - cervical/spasmodic torticollis
   - hand/writer's cramp
   - spasmodic dysphonia
   - blepharospasm

   Family members 18 years old or older can also join. Participants are asked to provide their medical and family histories and give a blood sample. Enrollment requires one 45-minute appointment at Massachusetts General Hospital. Those outside of the Boston area can enroll over the phone and by mail. Research test results will not be returned to participants.

2. Motor Learning in DYT1 Dystonia

Researchers seek to understand how changes in the DYT1 gene affect the ability to learn specific movements. Those who carry a mutation in the DYT1 gene (whether or not they have symptoms) as well as adult family members (who may or may not carry the mutation) are eligible to enroll Adults cannot take part if they:

- are less than 18 years old
- had brain surgery (including Deep Brain Stimulation)
- have dystonia symptoms that prevent completion of research tests.

Participants agree to a physical exam, videotaping, a blood draw and a series of "motor learning" tests (similar to video games). Participants also provide medical and family history information. Participation requires appointments at Massachusetts General Hospital and Massachusetts Institute of Technology over two and a half consecutive days. Travel and hotel costs will be covered for those outside of the Boston area. Participants who complete the study will receive $75. Participants will not learn the results of their testing.

Contact: Nutan Sharma, MD, PhD, Principal Investigator, (617) 724-9234, nsharma@partners.org; Trisha Multhaupt-Buell, MS, Research Coordinator, (617) 726-5470, tmulthaupt@partners.org

Public Eye

Media Watch

By Roxanne Ruzicka Maas, MS, CGC

March 24, 2008 – *MSNBC*, "When a Baby is Destined to Die"
Cheri Schoonveld is quoted and NSGC is mentioned in this touching article about how perinatal hospice programs help families facing a terminal prenatal diagnosis. The article covers a couple's decision to continue a pregnancy with trisomy 18 and the beautiful 29-day life of their baby. Speaking from her experience, Cheri said, "I think most people who have continued want to experience as much time with the baby as possible and want to hopefully meet the baby."

April, 2008 – *Your Health* magazine, "Tempting Flecks of Fate"
Robbin Palmer was interviewed for an article on the role of genetic counselors in assessing hereditary risk and interpreting genetic test results.

May 6, 2008 – *CBS Evening News with Katie Couric*
Anna Leininger was featured in a segment highlighting the process of genetic testing, the role of genetic counseling and the importance of knowing one's family health history. Two of Anna's patients, a brother and a sister who both tested positive for HNPCC were interviewed.

March 31, 2008 – *CBS News*, "Live at 9," Memphis, TN
Kami Wolfe Schneider appeared on this local television station's morning news show, discussing direct–to-consumer genetic testing and the importance of genetic counseling.

April 14, 2008 – *Fox News*, Memphis, TN
Eric Fowler was interviewed for this television broadcast to discuss genetic testing and counseling.

April 24-27, 2008 – *PBS*, "Smart Medicine"
Recurring several times, this local television program included a segment with **Kami Wolfe Schneider** discussing the opening of the new high-risk breast center at the Baptist Centers for Cancer Care in Memphis, TN and the role of genetic counseling.

June, 2008 - "Tracing Cancer Connections," *Heal: Living Well After Cancer* magazine
The cover stories of this summer issue opens with a women who decides to get a colonoscopy to address the colon cancer in her family after her beloved dog dies from colon cancer. This leads to the identification of an APC gene mutation and her foray into the world of inherited cancer predisposition and genetic testing. The extensive article reviews various forms of inherited cancer, including known and modifier genes, and the

process of genetic counseling to assess family history, arrange testing and help advise on medical management and testing for family members. Genetic counselor, **Rob Resta**, is quoted.

June, 2008 - "A Difficult Inheritance," *Heal: Living Well After Cancer* magazine
As a companion article to the above cover story, the Editor of this publication recounts her breast cancer diagnosis when her daughter is one year old, and her process of recognizing the cancer pattern in her family and ultimate decision to have genetic testing when she develops a recurrence as her daughter graduates from college. She describes her process of genetic counseling with **Becky Althaus**, including a diagram of her pedigree and the calculations of determining her risk for carrying a BRCA mutation. When she is found not to carry a mutation, she is pleased to be negative but also notes the value for her daughter in continuing to monitor the potential cancer risk that remains in her family. A link to the National Cancer Institute is provided for readers to find a local genetic counselor.

Send media items to Roxanne at rruzicka@gmail.com.

Public Relations Update

Three Questions Men Should Ask About Prostate Cancer

As part of NSGC's Prostate Cancer Awareness Month efforts and in honor of Father's Day, our PR firm, Public Communications Inc. (PCI), developed a media release recommending three questions men should ask about prostate cancer. Cancer SIG Co-Chairs, Joy Larsen Haidle and Rebecca Nagy, assisted PCI in developing these recommendations. During June, the news release was "pitched" to the media nation-wide.

We encourage you to contact your local media about genetic counseling and testing, too. The prostate cancer recommendations are posted at http://www.nsgc.org/client_files/news/053008_Prostate_Release.pdf and are available for your reference.

Be a PR Representative for NSGC

Mail or email the three questions release to the health or medical reporters at the following media outlets: hospital newsletters, local newspapers, local radio stations, local television stations, local/regional magazines and community group newsletters. Call each local media outlet to ensure that you are forwarding the information to the correct person.

Include a brief, personalized cover note with the reporter's name and title and a sentence or two explaining why you feel this information will be of interest to the reporter's viewers/readers. Follow-up with a phone call to ensure that the information has been received and to ask if the reporter has additional questions. If an article appears, contact NSGC and let us know – by working together, we can achieve more.