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President's Beat

Looking Forward with a Long-Term Plan

Greetings! It's a pleasure to take this opportunity to introduce myself as your 2011 President of the NSGC. I graduated from the Genetic Counseling Program at the University of Pittsburgh in 1998. Since then I have been employed by the University of Utah Department of Pediatrics, providing genetic counseling in pediatrics general genetics and specialty clinics. My volunteer experiences in the NSGC began as a member of the Pediatrics Special Interest Group (SIG), included a position on the Board of Directors from 2003–2005 as the Region V Representative, then as a member of several Committees, and now as President. As President, it is a privilege to be the spokesperson for our organization as we forge ahead in an exciting and challenging time for healthcare and genetics.

One of the first of many important items on the Board of Directors' agenda this year is the development of a long-term strategic plan for the NSGC. This long-term plan is necessary to ensure we are proactive in meeting the diverse needs of our members, our target audience of physicians and healthcare providers, and our patient population. Through an environmental scanning process we are seeking input from many and various stakeholders in genetics, healthcare and industry to help inform our discussion. We have asked these individuals to share their specific thoughts on where they believe genetics is going, and what they think it will look like in the future.

Our next step in developing this plan is a facilitated strategic planning meeting involving our Board, representatives from the American Board of Genetic Counseling (ABGC), and a representative from the Association of Genetic Counseling Program Directors (AGCPD). This meeting will be held from February 25-26, 2011 in Chicago. During this meeting, participants will identify the broad goals for the NSGC in preparing to meet the future. These goals will be developed into a long-term strategic plan for the NSGC that will help outline a direction for our future and can be used in all of our strategic and programmatic planning efforts.

During our meeting, the Board and participants will discuss and examine challenging questions such as, "What are the important issues facing the genetic counseling profession in the next several years?", "What should the NSGC become to help genetic counselors prepare for this future?" and, "What important goals will we set for ourselves

and the organization, and what paths shall we take to reach them?" This promises to be a thought-provoking and lively discussion as we consider the needs of our members in relation to the future of genetics in healthcare and society. We must ensure we are promoting the value of genetic counselors, while focusing our strategic and operational planning in the context of our collective future.

In addition to the long-term strategic planning meeting, many other important activities are taking place in the NSGC. Our Committees are busy embracing the many charges bestowed upon them by the Board of Directors this year. The Public Policy Committee is creating and revising position statements on Disability, Direct to Consumer Genetic Testing, Nondiscrimination, and Healthcare Reform. In addition, they are supporting the pursuit of licensure in several states and collecting supportive documents to maintain a coalition in support of the NSGC's Federal bill, to be introduced this spring. The Access and Services Delivery Committee is putting the final touches on a payer toolkit that members will utilize to compel regional and local payers to cover and reimburse genetic counseling services. They are also working diligently on several Practice Guidelines that address important trends and practices in our profession.

The Communications Committee is continuing its efforts to revise and enhance content for our new Web site by developing Web site content guidelines. Additionally, they are continuing to incorporate our brand message into all of our communications. The Annual Education Conference (AEC) Subcommittee of the Education Committee has been extremely busy reviewing the multiple outstanding proposals for educational content at the upcoming AEC in San Diego. The Education Committee is selecting educational content for webinars this year, developing the 2011 online course, and working on a plan for outreach education that will take place in 2012. The Membership Committee's activities this year include an overhaul of the NSGC Mentorship Program to make it a more utilized and beneficial program to our members. They are also administering the student rotation opportunities you have seen announced on the listserv, and reviewing the Board of Directors nominations process.

Our SIGs have also been incredibly active this year in submitting educational proposals to the AEC Subcommittee related to trends in their areas of specialization or interests. They are working diligently to create updated and informative content to be put on our new Web site for referring physicians and other healthcare providers, all consistent with our brand.

The NSGC is *your* professional organization and we encourage you to get involved! We are striving to make certain that genetic counselors become and remain not an adjunct, but integral part of the healthcare team. We have an exciting year ahead of us as we look to the future with a long-term plan for genetic counselors, the genetic counseling profession, and the NSGC's role in preparing us for that future.

Karin M. Dent, MS, LCGC
2011 NSGC President

A Pro/Con Discussion About the NSGC Nomination Process

***Editors' Note**: After receiving questions and comments from members about the NSGC's current nomination process for the Board of Directors, we felt it would be beneficial to offer readers a pro/con historical discussion about this process involving viewpoints from former NSGC Presidents. Their viewpoints are listed below, in order from most to least recent President.*

From both sides of the fence: Candidate and Nominating Committee member

By Elizabeth (Liz) Kearney, MS, CGC, MBA, NSGC Immediate Past President

One of the responsibilities of the NSGC's Past President is to serve as Chair of the Nominating Committee, a committee formed annually and comprised of both Board members and members-at-large, to select a slate of candidates for the following year's Board of Directors. As the first President elected under the NSGC's new Board election process, launched in 2008 to select the 2009 Board, it feels as though the process has come full circle. Now, in 2011, I begin my term as Chair of the Nominating Committee. It certainly seems appropriate to reflect on the successes and challenges of the new system, and I am happy to provide one perspective.

I learned about the NSGC's new Board election process in a manner similar to how other potential leaders might hear about it – I asked an expert. I mentioned to my friend **Kelly Ormond**, one of the NSGC's Past Presidents, that I was thinking of submitting my name as a candidate for President. I had previously served with Kelly on the Board, when I was the Region VI (West coast) Regional Representative.

Fortunately for me, Kelly was very encouraging and mentioned, "Are you aware of the new process for selecting Board members?" I had some knowledge that the NSGC had experienced governance changes, but I wasn't confident of the specifics. When I asked her to elaborate, Kelly explained that the NSGC had decided to heed the trend of other professional associations such as the National Association of Social Workers and the American Society of Association Executives, by adopting a new election process. Instead of having a "contested" election, where one candidate ran against another, the NSGC was moving to a slate election meaning the Nominating Committee would select a group of people to put forward to the membership for ratification. The advantages, she explained, were that:

1) The Nominating Committee could ensure a balance of backgrounds on the Board (e.g., geographical representation, practice area, years of experience, ethnic and gender diversity, employers) to better represent the NSGC membership as a whole

2) Members who "lost" elections were not publicly defeated and, therefore, good candidates were more likely to try again in the future

So far, the new process sounded pretty good to me. I always felt like my election to Regional Representative could have been a little biased because I worked for Kaiser Permanente in northern California at the time, so I probably got a pretty high percentage of those votes. My opponent was an excellent candidate herself, and would have made an outstanding representative.

Kelly went on to explain that members could nominate other members (or themselves) for consideration by the Nominating Committee. Each candidate who wanted to be considered would need to write short-answer essays to five to seven questions and go through a phone interview with a member of the Nominating Committee. Examples of the essay questions are:

- What is your vision for the NSGC and the genetic counseling profession?
- What is motivating you to seek a position on the NSGC Board of Directors?
- In your review of the current Strategic Plan, describe how your skills and experiences might contribute to the Board's implementation of the plan. In

crafting your answer, you can address specific initiatives or talk more broadly about the plan as a whole.
- Please highlight any skills or experiences you have, such as writing, communication, leadership, financial, or other skills that may not be evident in your CV.
- The NSGC Board of Directors values diversity of all kinds, including diversity in skill, specialty, experience, culture, ethnicity and gender. What diverse characteristics will you bring to the NSGC Board of Directors?

At first, I hesitated because the application process sounded pretty involved. It reminded of applications for graduate school! However, as I thought more about it, I realized it was an opportunity to demonstrate to a small group of engaged Committee members why I wanted to serve as President and why I felt I had the right skills for the NSGC's needs in 2010. When I ran for Regional Representative in 2002, I wrote short answers to two or three questions that were distributed to the membership with the ballots. While a few people did comment to me afterward that they liked my responses, most voted for me because they knew me personally.

Well, the new process was definitely more involved! I spent hours preparing my answers to the essay questions, which challenged me to think about my skill set, the timing, and commitment of serving as the NSGC's President. Several weeks later, I was interviewed by then-President **Angela Trepanier**, who grilled me with some pretty tough questions about my answers to the essays, as well as some unexpected questions about how I would handle difficult situations.

By the end of the evaluation process I was tired, but I also felt I had had the chance to explain my skills and be seriously considered for the role. I knew the Nominating Committee must have learned a lot about me because the process was very similar to a job interview. Angie had also asked me during the interview whether there were other leadership roles I might seek. I told her that indeed, if I were not selected as President, I was seriously considering a role as Chair of the Industry SIG. So, through the Board Election process, I was fairly confident I would find some type of leadership position because the Nominating Committee would definitely know that I was interested in contributing to the NSGC. .

Obviously, my story has a happy ending in that I was fortunate enough to be selected for President. I've had the subsequent gift of serving with the Board members selected for the slate, who are highly qualified individuals with diverse backgrounds. The NSGC would not have been able to achieve so many of its goals over the last two years without such a well functioning, qualified Board. I truly valued the many different perspectives that the Board members brought to each of our discussions. It helped me (and I believe everyone on the Board) think more broadly and consider many different viewpoints in every major decision.

I recognize that there are some drawbacks to the new system. No system is perfect. A slate election doesn't give the membership that final choice between two candidates. However, I know from conversations with colleagues that, most of the time, they didn't know how to select a candidate and professional associations have typically low (around 20%) participation in voting. In my opinion, a highly engaged Nominating Committee is

better able to seek the qualified individuals to create a balanced group than an entire membership. However, whether we are using the current process or another process to nominate and elect our Board members, improvements are always possible. Therefore, we should continue our commitment to evaluate the Board election process each year, and consider the benefits and drawbacks of other options – with the focus always being on the end goal of selecting a qualified, diverse and engaged Board of Directors for the NSGC.

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NSGC Elections: A Time to Decide, Not Just Ratify

By Wendy R. Uhlmann, MS, CGC, NSGC Past President 1999-2000

In 2007, the NSGC made significant changes for governance and elections. Reducing the size of the Board from nineteen to twelve members was a significant positive change. Having a smaller Board increases efficiency, ease of decision-making, and timeliness of response to issues. However, changing the way our elections are conducted has had the negative outcomes of limiting the role of the NSGC's members in candidate selection and has resulted in a significant decrease in the number of members voting. No longer are members given the opportunity to vote for candidates; instead, members are presented with a pre-selected candidate for each position and asked to ratify the Board slate.

Genetic counselors, including Past Presidents and Board members have remarked to me, "Why take the time to vote, it has already been decided." The result: far fewer NSGC members are now voting in elections, while the percentage of genetic counselors voting in the American Board of Genetic Counseling's (ABGC) elections has remained the same. In 2007, the last election the NSGC held where members were given a choice of candidates, 26% of eligible members voted, as compared to 16% in 2008 and only 14% in 2010. During this same time period, 33-34% of ABGC members voted in Board elections. Prior to changing our election process, NSGC member participation in elections had been on par with other professional genetics organizations (Table 1).

Another concerning change to the election process is the new composition of the Nominating Committee (NC). The NC is now almost the same entity as the Board as five of the seven members are current Board members; the Immediate Past President appoints

the two non-Board NC members. Three of these Board members will serve on the NC for three years with their presidential rotation, and potentially longer if appointed to the NC in a previous Board position. Non-Board members are truly a minority on the NC, which presumably puts them in a challenging position to advocate for nominees if Board NC members have a different perspective. The NSGC's Board is essentially in a position to select their successors.

Other genetics professional organizations, with the exception of the American College of Medical Genetics (ACMG), have either one or no current Board members on their NC, and stipulate that members cannot have served previously on their NC for at least three years (Table 1). Therefore, unlike other genetics professional organizations, the NSGC's NC is not "at arm's length" from the Board, which is problematic. For the reasons enumerated above and so the Board does not become insular, I think it is critical that the NSGC re-examine the composition of the NC and decrease the number of Board NC members.

In the letter that 2007 NSGC President **Cathy Wicklund** sent to the NSGC's members announcing changes to the Board and elections process, the point was made that there would now be a greater emphasis placed on membership input with the nomination process.[1] NSGC members have always been encouraged to participate in the nomination process and even consider nominating themselves. However, having a voice in the nomination process does not have the same impact as having a voice in an election. Submitting the names of nominees, who may not even be placed on the ballot, is quite different from voting directly for candidates who will serve in leadership roles and represent our professional organization.

One of the arguments made in support of the new election process is that "it helps reduce the attrition that can occur when strong volunteers decide not to run for office because of a previous lost election."[2] The fact remains that there will be genetic counselors who are not selected for the ballot who will be disappointed and may decrease their involvement; the new election process does not eliminate this possibility. In fact, some nominees, especially those not selected for the ballot year after year, may feel frustrated that a small group is making the decision and the wider membership is not given the chance to weigh in by casting their votes.

In the 2007 letter sent to the NSGC's members, the point was made that the new election process "…promotes the concept that it is the composite skills of all Board members, not just an individual, which makes the strongest Board. By offering a slate of candidates, the Nominating Committee is able to make sure that the best group of people will be representing NSGC any given year."[1] Clearly, there are desired skills, expertise and experience needed for a board to be successful, which the Board can communicate and the NC can take into consideration when selecting candidates. Board members can also submit nominees, just like other NSGC members. With the current rigorous vetting of nominees through an application and phone interviews, the NC should be able to put together a ballot in which any of the candidates has the ability to serve the organization well.

Generally, in well-established organizations only a small proportion of the membership is knowledgeable about Board activities, the key issues the organization is facing, and the

leadership needed to successfully run the organization. Similar to other genetics professional organizations, it seems reasonable that those involved in leadership roles and most active in the organization should name the President and potentially other officers as well. The "Directors at Large" should be elected by the members, as the term implies they should be. Reading candidate statements and selecting "Directors at Large" is a reasonable time investment and would make the membership feel that their votes matter. An election process that involves a combination of appointed and elected Board positions would both provide the NSGC with strong leaders and give members a voice in candidate selection.

The abilities to seek information, facilitate decision-making, and utilize critical thinking skills are central to our work as genetic counselors. Surely we can use these same skills to critically assess candidates to lead our professional organization. Genetic counselors are able to cast their vote and select the genetic counselors that serve on our ABGC Board. Certainly genetic counselors are just as capable to select Board members for the NSGC.

[1]President Cathy Wicklund, MS, CGC, August 31, 2007.
[2]President Cathy Wicklund's "President's Beat," *Perspectives in Genetic Counseling*, p. 1-3, Summer 2007.

Table 1: Genetics Professional Organizations: Election Process for the Board of Directors

	National Society of Genetic Counselors (NSGC)[1]	American Board of Genetic Counseling (ABGC)[2]	American College of Medical Genetics (ACMG)[3]	American Society of Human Genetics (ASHG)[4]
Board of Directors				
# members	12	10	18	17
Ballot	- Single candidate for each position. Members ratify slate of candidates.	- 4 candidates for Director positions (vote for 2) - Board elects officers	-1 candidate for President-Elect - 2 nominees for each Director position - Board elects Secretary, Vice President for Clinical Genetics, Vice President for Laboratory Genetics and elects or appoints Treasurer	- 1 candidate for President-elect - 6 candidates for Director positions (vote for 3) - Board elects Secretary and Treasurer, not same year
Nominating Committee				
# members	7	5	6	7
members	- Immediate Past President, President, President Elect, 2 Directors-At-Large and 2 non-Board members -Immediate Past President is Chair - Chair appoints non-Board members	- 1 member of the Board of Directors and 4 non-Board members who are certified members (diplomates) - 4 of the five members shall not have served on the Nominating Committee during the previous 6 years - Chair shall have served on the Nominating Committee the previous year.	- President, Past President, Secretary and 3 non-Board members - Non-Board members appointed by the President, usually in different genetics areas (e.g. Laboratory, Clinical) - Secretary serves as Chair - To date, all non-board members have served only one term on the Nominating Committee	- Members cannot be on current Board of Directors - 6 of the members cannot have served on Nominating Committee in past 3 years - 1 member should have served on Nominating Committee previous year - 1 member should have served on Board previously - President appoints Chair
Percentage of Eligible Members Participating in Elections				
Years	2010 – 14% 2009 – 21% 2008 – 16% 2007 – 26%	2010 – 34% 2009 – 34% 2008 – 33% 2007 – 27%	2010 – 41% 2008 – 33% Elections held every 2 years.	2010 – 32% 2009 – 33% 2008 – 26% 2007 – 26%
Year Electronic Voting Initiated				
Year	2005	2008	2004	2004

[1] Information obtained from NSGC - Meghan Carey, Executive Director.
[2] Information obtained from ABGC – Sheila O'Neal, Executive Director and Beth Crowley, Association Manager
[3] Information obtained from ACMG – Melissa Forburger, Director of Administration and Judith Benkendorf, MS, CGC, Special Assistant to the Director
[4] Information obtained from ASHG - Joann Boughman, PhD, Executive Vice President

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From new to old and old to new: The 2010 Nominating Committee experience from a veteran counselor's perspective

By Elizabeth (Betsy) Gettig, MS, CGC, NSGC Past President 1992-1993

When the NSGC approached me to serve on the 2010 Nominating Committee (NC) I was a little surprised [Read: Betsy is old and the NC process is new]. I have not been particularly active in the NSGC's affairs since our shift from a member-driven organization to a Board-driven model and, frankly, I have been on the sidelines observing the transition. I first hesitated and then agreed to serve having heard some concerns about a pre-set slate of candidates being less than ideal for our organization. Happily, what I found in the process was a thoughtful, well researched and much more open procedure than I expected.

To give a little context, my first role with the NSGC in the '80s was working on various aspects of the Annual Education Conference. I still believe this to be an excellent entry point to our professional organization, and had Special Interest Groups (SIGs) existed back then, I would have offered up my services to those endeavors as well.

I have forgotten the year – maybe 1983 [Read: Betsy was new then] – but my first Board type of NSGC job was when **Ginny Corson** was Past President II of the NSGC, and with that job you also had the role of NC Chair. That was my "Why me?" moment, but I think the reason Ginny approached me was that I was in Region 3 (the South), and Region 3 needed more involvement with the Board beyond the Regional Representative position. (Note: Regional Representatives are no longer part of the NSGC Board, but had the role of representing the different regional variations in genetic clinical practice. "At-Large" Board members are now elected, but the current NC considers the regional distribution of the entire Board in the nomination process). So back then I said yes to Ginny, thinking I would learn about the leadership process, which I did.

The former NC's work was fairly straightforward. Nominations were taken from the membership with a form (you wrote a paragraph saying why you nominated that person). The NC could add its own nominees, and everyone was ranked. I think the NC Chair only called the top two ranked people for each position, and if anyone declined the next ranked person was contacted. If they said yes, they wrote a statement to the membership to be mailed with ballots. If they said no, their name was forwarded to the next NC for consideration again. Two people went on the slate for each open office. The NC approved the final slate, the Board was notified and the ballot mailed to NSGC members. I don't recall the bylaws allowing the Board to circumvent the NC.

In the 1990s, **Wendy Uhlmann** and I both served as NSGC Presidents; we therefore got the NC Chair role as Past Presidents. There were discussions about formalizing the NC process and the 2000-2001 NC, which Wendy chaired, gets all the credit here. For example, they initiated the process of contacting nominees and requesting their CVs for review prior to selecting them for the ballot. Then, each NC member was responsible for contacting selected nominees for interviews. The process was multistep and still resulted in two candidates being selected for each office. I liked the detail, but sometimes found it too detailed; I later found out I was wrong. Wendy and her NC's contributions strengthened the nominating process.

When the restructuring of the NSGC's Board governance occurred, there were a number of changes in the nominating process. The first big change was listing only <u>one</u> candidate for each of the open positions. This still does not sit well with me. It reminds me of the kids at soccer who all get ribbons; face it, there are winners and losers in life and sometimes running and losing an office makes a candidate better. But we have one person per office, and that's the way it is for now.

The strengths and weakness of the NC are intertwined. The NC is top heavy in current presidential leadership. The current President, Past President and President Elect are on the NC. As current active Board members, these folks understand and support the mission of the NSGC Board, are engaged in strategic planning, can think about the nominating process in an ongoing manner (not just once a year), and can ensure which Board committees are active. They are familiar with current projects and activities of the Board. That said, you also have to consider that the Board's focus might not extend completely to the full membership, or reflect a degree of flexibility. The Board could become focused on the day-to-day versus big picture of the profession. There is also the potential for a lack of confidentiality, which might inhibit candid discussion of issues resulting in the absence of diversity of opinions, race, gender, age, geographic representation and tenure on the Board.

What I saw and heard in the NC process was positive. The NC did consider diversity and valued it. There was attention given to how long people had been on the Board, age, gender/ethnicity, and geographic representation.

A few comments stand out to me, which have both positive and negative potential. There were several remarks made about the "culture" of the Board – there was even a fact sheet on this topic given to NC members [Read: In my opinion, overkill]. Comments arose about how a nominee might "fit" into the current Board. Though a reasonable concern, a

strong Board can tolerate and foster good debate. You need strong opinions and risk takers to lead, and you want to go forward. I had to go to official government "school" when I was elected to public office in my community, and one class was about how to manage a Board. The lecturer said, "As elected public officials, you do not have to like the person seated next you on the Board, you simply have to work with them." That comment was so true during the next four years in my community work that I may have it tattooed on my body at some point! But I think for the NC this is a non-issue. Take talented people on, and it all works out.

When I was the NSGC's President there were many Board members, and about a third were Board spots the President appointed. I recall appointing **Vivian Weinblatt** to her first NSGC Board spot. Her first question to me after I made the invitation to join the Board was "Why me? You don't know me; we have never met." Well that's the point – you have to trust. You can't know every NSGC member, and you have to move outside your comfort zone and simply pick the best people for the job. I asked friends, colleagues, current and past NSGC Board members and Committee Chairs to pass names along to me. I was only interested in strong leaders and creative, critical thinkers. I did not know Vivian, but I got it right when I chose her for the Board.

I did not realize the number of appointments that are non-Board level positions like Committee Chairs. Committee Chairs used to be on the Board. When the NC catalogued all the openings among the Committee and Subcommittees, there were several. This information was presented late in the process to the NC. The reason to mention this is that an excess of candidates were nominated for leadership positions to the Board, and the nominees were highly skilled individuals. In the end, the NC discussed those not selected for the slated positions and considered many of these individuals for Committee or Subcommittee leadership positions. It was an excellent way to further cultivate the expertise of our membership.

In summary, the current NC experience impressed me by allowing every person on the NC to have his or her opinions heard and acknowledged. The misinformation I had was that the Board could overrule the process after receiving the NC recommendations. That may technically be allowed in the Bylaws, but has never happened. The Board took the NC's recommendation of the slate as is. The NC was deliberate and methodical about its charge, and the overall well being of the NSGC was the focus of our efforts.

First Annual "Genetic Counselling Awareness Week in Canada": A Great Success

By Allison Janson, MS, CGC, CCGC, Corissa Manou, MS, CGC, CCGC, and Jeanette Wilkins, MSc, CCGC, 2010 CAGC Genetic Counselling Awareness Week Subcommittee Co-Chairs

In 2010, the Canadian Association of Genetic Counsellors (CAGC) celebrated its twentieth anniversary. To mark this milestone, the CAGC Media and Communications Committee worked to develop and promote an annual "Genetic Counselling Awareness

Week." Genetic counselors across Canada embraced the week as an event to promote the profession and genetics to other healthcare providers and the community.

The inaugural Genetic Counselling Awareness Week took place from November 21-27, 2010 with a plan to recur annually during the fourth week of November. The week was a great success, and events celebrating and highlighting the genetic counseling profession took place across Canada. Canadian genetic counselors showed creativity and resourcefulness in organizing events and promotional materials, and many centers held multiple events throughout the week. The theme of Genetic Counselling Awareness Week for 2010 was 'Family History.' *Here are highlights from the week at various centers across Canada, from east to west:*

Several information booths were scattered across the East coast at universities and hospitals in Nova Scotia and Newfoundland, inviting individuals who passed by the opportunity to chat with a genetic counselor, grab some information and a snack.

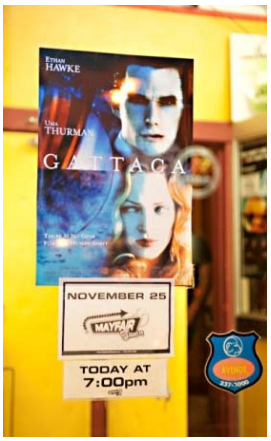
In Québec, Montréal's McGill University genetic counseling students took part in the festivities by setting up information booths and discussing the genetic counseling profession with passersby. In addition, two Québec-based genetic counselors, **Nathalie Bolduc** (President of the Association des Conseillers et Conseillères en Génétique du Québec) and **Gail Ouellette** (Director of the Regroupement Québécois des Maladies Orphelines) took part in a radio interview to promote genetic counseling and the Week.

McGill University genetic counseling students Justin and Alex (L), and Lynn and François (R) manning their information booth in Montréal

Ontario was also quite busy, as numerous centers held events. The genetic counselors at North York General Hospital expressed their creative talents by making a YouTube parody film entitled "What is a Genetic Counsellor." This four-minute humorous video was circulated online and had over 4,000 views!

Our Ottawa colleagues held a very successful screening of the movie "GATTACA," sponsored by a local art company called DNA11. This group took advantage of the opportunity to interact with interested members of the public by hosting a career fair prior to the screening.

With a visitor at Ottawa's Career Fair



"GATTACA" screening at DNA11 in Ottawa

Genetic counselors in Hamilton had a successful screening of the documentary film "In the Family" about hereditary breast-ovarian cancer – there is even a waiting list for future film screenings.

Genetic counselors from University Health Network and Mount Sinai Hospital in Toronto held an event (sponsored by GeneDx Laboratories) at a local pub entitled **"**Gene Scene: A parlor-style discussion on the Impact of Genetic Testing**"** and a lunch-and-learn information session for hospital staff about "Genetic Counselling in Clinical Practice." The Hospital for Sick Children's genetic counselors in Toronto organized and hosted Genetics Grand Rounds and set up an information booth and poster session in the hospital lobby. Elsewhere in Ontario, Kingston General Hospital also held hospital informational events, and the group in Sudbury participated by setting up hospital information booths.

In Winnipeg, Manitoba, events were planned throughout the week, including a film screening of "Twisted," arranged in collaboration with Dystonia Medical Research Foundation Canada, which had almost one hundred attendees. In addition, genetic counselors organized a book display at several bookstores. Readings from a selection of titles exploring families' experiences with genetic counseling and living with a genetic condition took place at a local café as part of "Java and Genes Coffee House." Furthermore, genetic counselors set up information booths in multiple hospitals and universities in the Winnipeg area, with the hopes to expand events to centers outside of the city during Genetic Counselling Awareness Week 2011.

Claudia Carilles at "Java and Genes Coffee House" at Ellice Cafe and

Information booth at St. Boniface College in Winnipeg

Theatre in Winnipeg

In parts of rural Alberta, Genetic Counselling Awareness Week 2010 table talkers and posters were placed in hospital cafeterias, physician lounges, and office waiting rooms. These generated several calls to genetic counselors inquiring about the services we provide and availability of these services.

Edmonton genetic counselors held a film screening of the documentary "In the Family" at the city's "Science in the Cinema" series, which partners with The Alberta Heritage Foundation for Medical Research Endowment Fund and Alberta Innovates - Health Solutions. Genetic counselor **Cynthia Handford** led a discussion about the film and genetic counseling with audience members after the screening. The Edmonton genetic counselors also gave various lectures to university and college students, and set up information booths at several regional conferences and in the lobby of the University of Alberta Hospital. The information booth included information on creating your own pedigree, a "Wheel of Genetic Counselling," genetics-themed electronic "Jeopardy" (complete with sound effects) and large fuzzy dice that illustrated inheritance patterns and common phenotypic traits.

Information booth at the University of Alberta Hospital in Edmonton

Kurston Doonanco manning the booth by the "Wheel of Genetic Counselling" in Edmonton

Calgary genetic counselors set up information booths and scattered table talkers and posters throughout local hospitals. They hosted a "Science Café" at the University of Calgary for the general public, which was well received. Furthermore, discussions took place with the Calgary Board of Education and Catholic School Board focused on the role of genetic counselors in assisting teachers with the development of genetics teaching tools for high school Biology curricula.

Saskatchewan and British Columbia also participated in the festivities by setting up information booths, presenting a lecture on hereditary cancer syndromes to the British Columbia Cancer Agency, and submitting publications to the Provincial Health Services newsletters.

As an incentive to participate and plan events, the CAGC offered a prize to the most creative and enthusiastic group. This was a gift certificate of $200 to a restaurant of the

winning group's choice. **Congratulations to the 2010 winners – the genetic counselors from Winnipeg, Manitoba**. Additionally, each participating center was entered in a random draw for a $50 gift certificate. **The winners of the draw were the genetic counselors from North York General Hospital in Toronto, Ontario.**

These are just some highlights from events that took place across Canada. Genetic counselors were extremely enthusiastic about this week, which was demonstrated by the amount of time they devoted to planning and executing these events. Groups reported a greater sense of camaraderie and teamwork as they planned and participated in their events together. GC Awareness Week also provided opportunities to generate excellent materials about genetic counseling and to network with others, both of which can be built upon to help grow the profession over time.

We'd like to acknowledge the Genetic Counselling Awareness Week Regional Coordinators for all their hard work: **Martha Balicki**, **Sajid Merchant**, **Jessica Hartley**, **Rachel Vanneste**, **Tina Babineau Sturk** and **Mary Connolly-Wilson**.

Thank you to all who participated and helped to make the week a success. We look forward to planning Genetic Counselling Awareness Week 2011!

For more detailed information about the events that took place, please check out the Genetic Counselling Awareness Week website at http://www.genetic counsellors.ca.

Extending Comprehensive Cancer Center Expertise in Clinical Cancer Genetics and Genomics to Community-Based Practices

By Kathleen R. Blazer EdD, MS, CGC, Julie Culver, MS, CGC and
Deborah J. MacDonald, PhD, APNG, City of Hope Comprehensive Cancer Center

***Editors' Note**: To avoid any possible conflicts of interest, the authors would like to disclose their affiliation with the organization highlighted in this article. The authors are employed by City of Hope, an independent medical organization designated by the National Cancer Institute as a Comprehensive Cancer Center. City of Hope offers grant-subsidized, fee-based courses to clinicians as a part of their care delivery model.*

The traditional academic model for the delivery of genetic cancer risk assessment (GCRA) involves one or more consultative sessions with an interdisciplinary team that may include genetic counselors, advanced practice nurses, one or more physicians (generally a medical geneticist or oncologist), and, in some settings, a mental health professional. However, most people receive their medical care in the community setting, where clinicians often have limited time, resources, or expertise to dedicate to the GCRA process.

Established in 1996, City of Hope Division of Clinical Cancer Genetics (CCG) in Duarte, California provides full-spectrum clinical GCRA services through its Cancer Screening &

Prevention Program Network (CSPPN), delivers professional education and training in cancer genetics through its Cancer Genetics Education Program, and supports prospective cancer genetics research initiatives through an Institutional Review Board-approved Hereditary Cancer Registry protocol.[1-6] This article describes how the CCG leverages its clinical, education, and research resources and expertise to promote the delivery of evidence-based GCRA services and cancer genetics research participation in the community setting.

Transition from the multidisciplinary academic health center model to community-based GCRA

As a major component of maintaining National Cancer Institute (NCI)-Comprehensive Cancer Center status, the CSPPN serves as a central clinical and research resource to a growing network of community-based medical centers and clinicians. Detailed descriptions of the establishment and growth of the CSPPN have been previously published. [1, 3] Community-based centers are contracted with City of Hope for program development, GCRA training, and ongoing practice-centered support to promote quality care. The program development activities are tailored to address the needs and resources of each community center. In addition to a thorough orientation to City of Hope GCRA protocols, advice and assistance is provided to each contracted affiliate regarding clinic and family history instruments and selection of pedigree database software.

Different models for different settings

The clinical satellite affiliates of the CSPPN are described in Table 1. To date, the CSPPN has provided comprehensive GCRA services to more than 7,000 individuals and their families, with approximately 20% of these stemming from our satellite affiliates. The continued growth and flexibility of the CSPPN is enabled by information technology. Videoconferencing facilitates the delivery of clinical services via telemedicine at some sites, and Web conferencing brings affiliates together with the interdisciplinary City of Hope team for cancer genetics case conferencing through participation in the CCG Working Group, a component of the CCG Community of Practice (described below).

Alternative modes of GCRA delivery enable cost-effective community medical center participation. The choice of models is dependent, in part, on the availability of qualified staff and the local institution's economic environment. Billing for mid-level services is sometimes possible through facility fees or individual provider codes. Some affiliates have helped justify under-reimbursed program costs by apprising administrators of potential downstream revenue from cancer screening, chemoprevention, and surgical risk reduction interventions.[3]

Quality assurance in GCRA: The Clinical Cancer Genetics (CCG) Community of Practice

CSPPN affiliates have ongoing access to the evidence-based updates and practice-centered support essential to sustaining an informed community-based GCRA practice through participation in a dynamic distance-mediated CCG Community of Practice. This is centered on two Continuing Medical Education (CME)-accredited Web conference activities:

(1) "Clinical Cancer Genetics (CCG) Working Group"
The CCG Working Group is an interdisciplinary cancer genetics case conference series conducted each week by City of Hope's clinical team. CSPPN and affiliated clinicians across the U.S. present cases from their community practices via Microsoft Live™ Web conference interface for discussion and recommendations on risk assessment, surveillance, risk management, and identification of research eligibility for cases covering the full spectrum of hereditary cancer.

(2) "Topics in Clinical Cancer Genetic Research"
Topics in Clinical Cancer Genetics is a weekly one-hour Web-conference seminar series focused on timely issues in clinical cancer genetics, cancer epidemiology and cancer genetics research, alternating among didactic lectures, case-based literature reviews, and basic research journal club. City of Hope faculty, guest lecturers from other academic institutions, CSPPN affiliates and alumni of City of Hope's "Intensive Course in Community Cancer Genetics" (described below) are included in the roster of presenters to ensure that the topics covered address the practice-centered learning needs of community-based participants.

Expanding the expertise of the academic health center: The Intensive Course in Community Cancer Genetics and Research Training

Interdisciplinary GCRA training and continuing Continuing Medical Education (CME)/Continuing Education Unit (CEU) activities are essential to extend the expertise and resources of the academic health center to the community-based setting. In response to the national need for specialized training in GCRA, the CCG Cancer Genetics Education Program has developed an NCI-funded (R25 CA112486) "Intensive Course in Community Cancer Genetics and Research Training" for community-based clinicians (genetic counselors, oncology or genetics-trained physicians and advanced practice nurses). The goals of the course are to increase the number of clinicians with practitioner-level competence in GCRA and to promote community-based research participation. These goals are accomplished through a three-phase CME/CEU-accredited program of distance didactic learning (Phase 1), interdisciplinary face-to-face training (Phase 2), and continuing professional development activities (Phase 3), to support the integration of high-quality, evidence-based GCRA services and research into practice.

To date, more than 220 clinicians representing community-based clinical practices in 45 U.S. states, Argentina, Brazil, Canada, Chile, Spain, and Hong Kong have completed the course. Upon completion of Phase 2 on-campus training, course participants are invited to join the roster of CSPPN affiliates and intensive course alumni who participate in the CCG Community of Practice for twelve months of professional development and case-

based support upon return to their practice settings (Phase 3). A number of CSPPN affiliate clinicians established their formal collaborations with City of Hope as a consequence of their participation in the course. Moreover, a growing roster of course alumni continue to participate in Phase 3 Community of Practice professional development activities well beyond the prescribed twelve-month period. Findings from an action research project conducted with course alumni revealed that many continue engaging in the Community of Practice as a source of ongoing support for interdisciplinary evidence-based GCRA patient care and research collaboration in their practices[7].

Summary

No matter what models are employed to address the demand for more efficient and broader coverage of GCRA services, no model should compromise informed decision making by patients, or appropriate application and interpretation of genetic and genomic test information. Through innovative clinician education, technology, and sustained collaboration, the comprehensive cancer center can play a significant role in extending evidenced-base genetic/genomic information and best practices in GCRA into the community setting. Our experience with continued demand for comprehensive GCRA training and extended participation in the CCG Community of Practice by a growing number of intensive course alumni demonstrates that quality care is important to a highly motivated subset of practitioners across practice disciplines and clinical settings.

Acknowledgements

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Table 1. Models of Practice within City of Hope's Cancer Screening & Prevention Program Network (CSPPN)

Program/Location	Delivery Model/Staff Composition
Academic Health Center	
City of Hope Medical Center Duarte, California Host Institution	• Initial visit (including genetics-focused physical exam) with board-certified genetic counselor (CGC) or advanced-practice nurse with genetics credentials (APN) and physician (onco-geneticist); follow-up/results disclosure visit with same cancer risk counselor and physician team or phone disclosure for straightforward cases. • Alternative two-visit model – initial visit with cancer risk counselor only; follow-up/results disclosure visit with same cancer risk counselor and physician (genetics-focused physical exam on follow-up visit)

	• Urgent slots held in scheduling template to accommodate patients seeking GCRA in context of diagnosis and treatment planning • Patients invited to participate in the Hereditary Cancer Genetics Registry • Cases reviewed during CCG Working Group**
CSPPN Community Hospital and Cancer Center Affiliates	
St. Jude Medical Center/ Virginia Crossen Cancer Center Fullerton, California	• Initial visit at Saint Jude with CSPPN APN credentialed in genetics; follow-up/results disclosure visit with same APN and physician (onco-geneticist) • Patients invited to participate in the Hereditary Cancer Genetics Registry • Cases reviewed during CCG Working Group**
Cancer Center of Santa Barbara Santa Barbara, California	• Program evolved from two-visit model of initial visit at Santa Barbara with CSPPN APN credentialed in genetics with follow-up/results disclosure visit at City of Hope with same APN and physician (onco-geneticist) to both visits delivered on-site at Santa Barbara by a CGC* • Patients invited to participate in the Hereditary Cancer Genetics Registry • Cases reviewed during CCG Working Group**
Good Samaritan Medical Center, Phoenix Arizona	• Program administered by an APN board certified in genetic counseling*, with clerical support and oversight by a local oncologist • Initial and follow-up/results disclosure visits conducted by the APN/CGC administrator or a CGC* • Patients invited to participate in the Hereditary Cancer Genetics Registry • Cases reviewed during CCG Working Group**
St. Joseph's Medical Center Orange County, California	• Program administered by a CGC*, with program support from a medical oncologist, surgical oncologist and a colorectal surgeon • Initial and follow-up/results disclosure visits by the CGC administrator or one of two additional CGCs • Patients invited to participate in the Hereditary Cancer Genetics Registry • Cases reviewed during CCG Working Group**
St. Alphonsus Medical Center Boise, Idaho	• Program administered by a CGC* • Initial and follow-up/results disclosure visits conducted by the CGC administrator • Patients invited to participate in the Hereditary Cancer Genetics Registry • Cases reviewed during CCG Working Group**
Underserved Outreach Affiliates	
Toiyabe Indian Health Project (Indian Health Service) Bishop, California	• Initial and follow-up/results disclosure visits conducted by a City Of Hope cancer risk CGC and physician via telemedicine (with RN at Toiyabe site for patient and tech support) • Patients invited to participate in the Hereditary Cancer Genetics Registry • Cases reviewed during CCG Working Group**
Los Angeles County Olive View/UCLA Medical Center Sylmar, California	• Grant support facilitated the establishment of this underserved minority outreach clinic in collaboration with a regional County hospital • Patient participation facilitated through the Hereditary Cancer Genetics Registry • Initial visit conducted at the Olive View campus by a Spanish-English speaking City of Hope CGC; follow-up/results disclosure visit conducted with same CGC on-campus at Olive View and onco-geneticist participating from City of Hope by telemedicine • Genetic analysis supported by compassionate funding (patient must meet NCCN criteria for *BRCA* testing) • Program is supported by chief of oncology and bilingual patient navigators at

	Olive View to facilitate risk-appropriate cancer screening and prevention services prescribed through the GCRA process • Cases reviewed during CCG Working Group**
* Received formal training in GCRA through City of Hope professional education initiatives ** The CCG Working Group is a weekly CME-accredited case discussion forum which can be accessed by Web conferencing. See text for description 1. Cancer Screening & Prevention Program (CSPPN) 2. Division of Clinical Cancer Genetics (CCG) 3. Certified genetic counselor (CGC) 4. Advanced practice nurse (APN)	

Table adapted from: MacDonald DJ, Blazer KR, Weitzel JN. Extending comprehensive cancer center expertise in clinical cancer genetics and genomics to diverse communities: The power of partnership. *Journal of the National Comprehensive Cancer Network.* 8(5):615-624. 2010.

References

1. MacDonald DJ, Sand S, Kass F, *et al*. The power of partnership: Extending comprehensive cancer center expertise in clinical cancer genetics to community breast care centers. *Seminars in Breast Disease.* 9:39-47. 2006.
2. Blazer KR, Grant M, Sand SR, *et al*. Development of a cancer genetics education program for clinicians. *J. Cancer Educ.* 17(2):69-73. Summer 2002.
3. MacDonald DJ, Blazer KR, Weitzel JN. Extending comprehensive cancer center expertise in clinical cancer genetics and genomics to diverse communities: The power of partnership. *J Natl Compr Canc Netw.*8(5):615-624. 2010.
4. Blazer KR, Grant M, Sand SR, MacDonald DJ, Uman GC, Weitzel JN. Effects of a cancer genetics education programme on clinicians knowledge and practice. *J. Med. Genet.* 41(7):518-522. 2004.
5. Blazer KR, MacDonald DJ, Ricker C, Sand S, Uman GC, Weitzel JN. Outcomes from intensive training in genetic cancer risk counseling for clinicians. *Genet. Med.* 7(1):40-47. 2005.
6. Blazer KR, MacDonald DJ, Justus KA, *et al*. Creating tomorrow's leaders in cancer prevention: A novel interdisciplinary career development program in cancer genetics research. *J. Cancer Educ.* 21(4):216-222. 2006.
7. Blazer KR, Sand S, Hilario J, Weitzel JN. Using action research to evaluate and innovate a program of personalized cancer genetics training for personalized medicine. Paper presented at AACE International Cancer Education Conference: The Art and Science of Cancer Education and Evaluation, Houston, Texas. 2009.

For Your Practice

The Value of Peer Support

By Natalie Witkin, Director, In Our Genes, Hereditary Breast and Ovarian Cancer Programs, Willow Breast Cancer Support Canada

When told they carry an inherited mutation that increases their risk for breast and ovarian cancer, many women are overwhelmed. After the initial shock of testing positive, they face tough choices from how to manage their risk to how to tell family members about their results. Where do they fit into the medical system if they are at risk but not diagnosed with cancer? Whom can they talk to if their breast cancer is inherited? Finding a group of women who share a similar experience and understand the impact of carrying a *BRCA1* or *BRCA2* mutation can be a tremendous help.

On October 24, 2009 in Toronto, Ontario, Daniela was told by her genetic counselor that she had tested positive for a *BRCA2* gene mutation. At the same time, Daniela's mother was dealing with her third bout of breast cancer and a recent ovarian cancer diagnosis. Paralyzed by fear and uncertainty, Daniela took two months off work to cope with her new reality. During this time she had the help of a genetic counselor who was empathic and skilled at explaining Daniela's risk factors and risk management options. She took time to explain complicated information (many times!), but it wasn't enough. Daniela desperately wanted to connect with another mutation carrier – someone who understood what she faced. She wanted to know how they coped and what they were doing to manage their cancer risk.

Daniela reached out to local breast cancer organizations, but they did not have any supportive services for at-risk *BRCA* mutation carriers. They welcomed her to attend a breast cancer support group, but Daniela didn't have cancer.

Where did she fit in? She tried talking to her family and friends, but found she was spending most of her time educating them on what it meant to be a *BRCA* mutation carrier, and why she was considering prophylactic surgery. Daniela was grateful that her husband, family and friends were supportive, but she didn't feel like they really "got it." In spite of everyone's best intentions, Daniela still felt isolated and alone.

Connecting with other gene mutation carriers can be challenging because the number of carriers is small. Those at risk may be hesitant to talk about their genetic condition for a number of reasons: explaining genetic disease is complex; disclosure of genetic information often directly impacts other family members; avoiding breach of confidentiality, since sharing personal genetic information can indirectly implicate other family members; concern over genetic discrimination; apprehension regarding the reaction of, or possible stigmatization by, family or friends[1].

Daniela could not find any local organizations that provided support and information to those who carry *BRCA* mutations. In desperation, Daniela posted a comment on an online message board and connected with a few other women like her. After corresponding a few times, they made arrangements to meet, and Daniela drove over two hours to meet them in a coffee shop. There she found tremendous comfort and support by being in the company of other women who shared her experience.

A friend suggested that Daniela call Willow Breast Cancer Support Canada (Willow), a Canadian national not-for-profit organization that provides information and support to anyone at risk or diagnosed with breast cancer. Willow understood that the needs of this community were different from those of breast cancer survivors. The Willow team met with Daniela and two other women who were at risk; their goals were to develop the tools and training needed to start a peer-led support group specifically for women who tested positive for a *BRCA* mutation. In September 2009, the first *BRCA*-positive support group met at Willow's facility in Toronto, Ontario.

When twenty-three women showed up that first night, Willow recognized there was a need for a comprehensive national, peer-led resource for women at risk or affected by hereditary breast and ovarian cancer (HBOC). This led to the establishment of *In Our Genes*, a program that provides support and information to high risk individuals and their families. *In Our Genes* helps the HBOC community understand and cope with the issues associated with a diagnosis and empowers them to make informed decisions.

Daniela Pereira, Valerie Cooper and Natalie Witkin celebrate the one-year anniversary of the Willow support group they founded

Willow serves anyone at risk for, or diagnosed with, breast cancer by providing personalized information and emotional, social and practical help, delivered by a *BRCA* mutation carrier who is trained in providing peer support and information. At the core of the peer support model is the understanding that nothing compares to getting support from others who have been through a similar experience. Peer support gives people the opportunity to be heard and encourages clients to be informed; peers also provide insight based on personal experience. This experiential approach empowers people by imparting knowledge in ways not typically offered by the medical system[2].

Peer support offers many real and lasting benefits, including:

- Reducing isolation by providing a sense of universalism or shared experience
- Reducing anxiety by offering practical advice and self-help skills for coping with difficult emotions

- Helping people develop effective strategies to cope with treatment-related problems[1]
- Providing the appropriate resources to better understand a diagnosis
- Discussing how to navigate the healthcare system and advocate on one's own behalf

Willow recognizes that a wide range of medical, psychosocial and community-based support services are required to fit the complex needs of individuals with HBOC. Genetic counselors assess a family medical history, determine eligibility for testing, analyze test results, help patients and families make sense of their situation, and inform patients about risk management options. An individual's cancer risk is managed through surveillance, chemoprevention or prophylactic surgeries. In high-risk centers or hospitals, psychosocial support to cope with the emotional effects of being at risk for HBOC may be offered by professionals who are often overburdened with treating those most distressed by their diagnoses. *In Our Genes* provides valuable peer support outside the hospital setting, and can offer unrestricted access and unlimited time to address the concerns of this underserved community.

Willow's free services available to those at risk include:

- **Peer support**: connects people with a *BRCA* mutation carrier or breast cancer survivor trained in providing peer support who understands the impact of a diagnosis and offers information, emotional support and encouragement
- **Information services**: offers personalized, current and credible information on all aspects of HBOC
- **Support Group Program**: gives those at risk the knowledge and tools necessary to start and sustain community-based support groups
- **Inourgenes.ca**: a dedicated website for the HBOC community providing quality resources and links to a vast number of reputable online sources and an e-newsletter about the latest research, personal interest articles and local events
- ***BRCA*+ Peer Support Groups**: gives women who are *BRCA*-positive the opportunity to meet as equals to give each other support on a reciprocal basis
- **willow-talk.org**: a safe online community to connect with others, exchange information and share experiences
- **Translation:** free interpreter services for individuals wishing to speak in their language of choice during a support call

Comprehensive cancer care is best provided through collaboration between the medical system and community-based organizations and programs. Willow is committed to working with the medical community to share the load. The organization recognizes that it is in a unique position to empower people affected by hereditary breast and ovarian cancer by imparting knowledge, sharing experience, assisting with healthcare navigation and, most importantly, taking time to listen and reassure them that they are not alone. Willow does not give medical advice, recommend doctors or hospitals, offer opinions or preferences, or make decisions for users of its services. Willow also does not approve or endorse any particular treatment or course of action.

In Our Genes values the expertise and experience of professionals working with high-risk individuals. Participating in workshops and conferences organized by organizations such as Canadian Association of Genetic Counsellors (CAGC), Canadian Association of Psychosocial Oncology (CAPO), and Facing Our Risk of Cancer Empowered (FORCE) has provided an opportunity for Willow to identify gaps in supportive care and challenges faced by health professionals. In response, *In Our Genes* has formed a partnership with genetic counselors to develop relevant and practical written resource materials, such as fact sheets to address some of the issues faced by those living at risk.

Willow is working with health care professionals to improve patient and physician knowledge about HBOC. **Willow needs your help to assemble a network of health professionals to share up-to-date information with the HBOC community.** If you are interested in lending your expertise and advice to the development of information and support programs, please contact *In Our Genes* to learn more.

Many high-risk clinics now offer their patients literature about *In Our Genes.* If you are interested in obtaining copies of our free material, please visit www.inourgenes.ca to download the PDF.

References

1. Genetic Alliance, The New York - Mid-Atlantic Consortium for Genetic and Newborn Screening Services. Understanding Genetics: A New York, Mid-Atlantic Guide for Patients and Health Professionals. 2009. Posted in the Genetic Alliance Resource Repository at http://www.resourcerepository.org/documents/1247/understandinggenetics:anewyork,mid-atlanticguideforpatientsandhealthprofessionals/#

2. Rutledge R, Robinson L. Community based organizations are critical partners in providing complete cancer care. *Current Oncol.* 16; 29-33. 2009.

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The HDBuzz Web site Launches

By Jeff Carroll, PhD and Deepti Babu, MS, CGC

2011 brought the launch of a new online resource that those working with families for Huntington disease (HD) genetic counseling will find helpful: HDBuzz (http://hdbuzz.net/). HDBuzz is a clearinghouse for high-quality research news about HD for the global community, written in understandable terms by clinicians and researchers who work on HD. While the stories are written by researchers, to avoid conflicts of interest these contributors will not report on their own work. The site highlights laboratory and clinical research, with the aim of helping those in the HD community understand the latest achievements in the world of medical and scientific research. Also look for in-depth coverage of HD scientific conferences and other events of interest to the community.

Dr. Ed Wild and Dr. Jeff Carroll, HD scientists in the U.K. and U.S.A., respectively, founded HDBuzz. A unique consortium of HD family support organizations funds the site, with the primary supporters being the Huntington's Disease Society of America, Huntington's Disease Association of England and Wales, and the Huntington Society of Canada. More global HD organizations may join the consortium in later 2011. No funding is accepted from pharmaceutical companies or organizations with a vested interest in a particular treatment or technology, and no financial contributor has editorial control of HDBuzz content. An oversight committee of independent clinicians, scientists and general public community members reviews all site content.

All HDBuzz content will be translated into several languages (Spanish currently and hopefully French, German, and Dutch in the near future). It is available via free syndication to other HD community web sites like www.hdsa.org, www.hda.org.uk, and www.huntingtonsociety.ca. HDBuzz has news feeds to several social networking sites

like Twitter, Facebook, and Google Buzz via HDBuzzfeed. Updates are available via email, and if you have a Web page or blog you can freely use content automatically via RSS feeds.

HDBuzz is the result of an exciting collaboration between many people all over the world, and we hope the content will reach as far as possible. HD knows no cultural boundaries, and neither should access to information about HD.

Do you have a topic or article on HD that you'd like to see written about on HDBuzz? Or do you have feedback or comments? Feel free to contact us at Editors@HDBuzz.net.

Licensure / Billing & Reimbursement

The Coding Corner

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

The Coding Corner is supported by the Coding Subcommittee of the 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 CPT/ICD coding in genetic services as well as keep the membership educated regarding billing and reimbursement issues.

Billing and Reimbursement: Updates, Ongoing Efforts, and a Call for Action

The NSGC's strategic initiatives for 2011 continue to strive for professional advancement of genetic counselors as individual healthcare providers. Over the past five years a lot of activity has occurred, and the NSGC and its members continue to make strides in raising the profile and stature of genetic counselors through federal, state, and local efforts in billing, reimbursement, and credentialing. Each of these efforts is critical to achieve health insurance plan recognition and reimbursement for our expertise and services. The future of the profession depends on our success! The following reviews the status of our efforts at the federal, state, and local levels and identifies opportunities for even more membership engagement. Our successes are the results of the persistent and dedicated work of a number of your colleagues and the NSGC staff; however, everyone has a role to play in the outcomes. These concerted and varied efforts will very likely have great impact on the future of our profession.

Achievements

- In 2005, genetic counselors gained the ability to apply for a National Provider Identifier (NPI), a standard unique health identifier for health care providers, in accordance with the Health Insurance Portability and Accountability Act

(HIPAA), and required by the Centers for Medicaid and Medicare Services (CMS) and most health plans in order to facilitate billing and reimbursement. Our ability to obtain an NPI begins to acknowledge that genetic counselors are unique providers. The ability to use an NPI for independent billing with Medicare will require Congress to act to include us as providers for Medicare. (See Ongoing Federal Efforts)

- In January 2007, the American Medical Association added a new Current Procedural Terminology (CPT) code for 'Medical Genetics and Genetic Counseling Service', 96040, for genetic counseling services provided by non-physician genetics providers only. According to the 2010 Professional Status Survey (PSS), 15% of genetic counselors report billing under their own name with this code. For more information on how to incorporate this billing code in to your clinical practice, we encourage you to participate in the NSGC's 2009 Online Coding Course, "Learn the Three C's to Maximize Your Service Delivery Model: Coding, Credentialing and Compliance" that is still available for Continuing Education Unit (CEU) credits.

- In 2010, genetic counselors were added to the Department of Labor's List of Standard Occupational Classification (SOC). Inclusion permits collection and dissemination of occupational data for federal statistics and demonstrates that the work of genetic counselors is sufficiently distinct from that of any other occupation.

- As of January 2011, ten states are issuing licenses, three states have passed bills and are in the process of rulemaking, and about twenty states are preparing to introduce bills. The primary purpose of licensure is to protect the public's health. Licensure sets the standards for qualified providers and prohibits unqualified individuals from acting as genetic counselors. Licensure will reassure consumers that individuals who provide genetic counseling are qualified to do so. Licensure should also help support efforts for reimbursement for genetic counseling services; however, licensure alone will not guarantee the ability to bill for these services.

Ongoing Efforts

- Although the above achievements offer some measure of federal recognition, the NSGC seeks clearer federal regulation from CMS defining who can bill Medicare for delivery of genetic counseling services. The goal of this federal effort is for genetic counselors to be recognized by CMS as independent providers, such that genetic counselors would be able to bill Medicare for their services directly. This would improve patient access to genetic counseling services and set an example

for private third party payers. The NSGC Public Policy Committee is working with members of Congress to draft such legislation; however, passage of new federal legislation can take many years to accomplish. We will notify the membership when your help is needed.

- The NSGC has entered into a collaborative partnership with Informed Medical Decisions to strategically educate the major national health insurance plans about the advantages of developing coverage policies for genetic counselors and our services. This will significantly improve access to quality genetic counseling services. These are highly coordinated and planned initiatives to ensure the highest benefit for all parties involved: genetic counselors as a profession, the payers, and the patients receiving care.

- The NSGC's public policy staff and experienced lobbyists continue to work with genetic counselors who are seeking state licensure. The NSGC has developed resources including model legislative language, guiding principles, and supportive documentation that explain the critical elements of state licensing legislation and ensure uniformity in the practice of genetic counseling of the highest quality. These details will help to ensure consistency between states and with federal legislation. The legislative process for issuing licenses, the legislation climate, and financial implications are typically unique to each state. The Licensure Subcommittee of the Access and Service Delivery Committee is a resource that all states should use in licensure efforts. The NSGC has licensure grants available to support the efforts of individual states towards licensure. For more information, contact John Richardson at the Executive Office at jrichardson@nsgc.org.

Get Educated, Get Involved

It is imperative that each member educates him or herself about these issues. The economic aspects of health care are gaining more scrutiny and we have a responsibility to become savvier to the business side of our practices. If we don't set the rules for how we run our practices, others will set them for us – which may not be optimal for us or, more importantly, for our patients.

The first step in making local progress towards reimbursement for your services is educating yourself. Sadly, according to the 2010 PSS 25% of genetic counselors are not billing for their services and 11% do not know how they bill for their services. Furthermore, 37% are unaware of whether there had been any changes to their reimbursement from 2008 to 2010. This is over one third of our reporting membership!

The Access and Service Delivery Committee continues to work hard to provide opportunities for you, our membership, to become informed. These include the NSGC's 2009 Online Coding Course "Learn the Three C's to Maximize Your Service Delivery Model: Coding, Credentialing and Compliance," the NSGC's "Tools for Your Practice," which includes a billing and reimbursement toolkit, and a forthcoming credentialing

online course. Each institution and state will have its own regulations and requirements, and genetic counselors can also learn immensely by meeting with their compliance and billing representatives, as well as hospital administrators and contracting departments. We all have a role to play in shaping our future.

"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*** *at shannagustafson@gmail.com or* ***John Richardson*** *at jrichardson@nsgc.org.*

SIG Speak

From the Public Health Special Interest Group

Primary care physicians and direct-to-consumer genetic testing

By Whitney Cogswell, MS

Thanks in part to the Public Health Special Interest Group (SIG), I was able to complete my capstone project at the University of North Carolina Greensboro. I knew early in my graduate training that I was interested in a project focused on primary care physicians' knowledge or awareness of genetics. When I presented this thought to my capstone committee, they suggested that we research physicians' awareness of direct-to-consumer (DTC) personal genome testing. With this being a new, relevant, and exciting topic, we decided to run with the idea.

For this study, DTC genetic testing was defined as genetic testing that scans an individual's entire genetic makeup for potential health risks and is marketed directly to consumers. With a number of companies open for business, there have been numerous concerns regarding this method of genetic testing. The literature reports concerns relating to clinical and analytical validity and utility, how test results are communicated, the use and cost of follow up, and potentially misleading claims made by some companies[1-3]. Two previous studies, one in Japan[4] and one in the US[5], found that a minority of healthcare providers are aware of DTC genetic testing, and even less have experience speaking to patients about it.

We focused our efforts regionally by surveying primary care physicians in North Carolina. The sample was limited to Family and Internal Medicine physicians because we felt these physicians were the most likely candidates to handle questions about DTC genetic testing. Our survey was designed to assess physician awareness, experience, opinions, and self-perceived preparedness regarding DTC genetic testing.

Out of 2,352 eligible participants, 382 (16.2%) responded. Awareness of DTC genetic testing was similar to that found in the literature. 38.7% of physicians surveyed were aware of DTC genetic testing. The most frequently cited sources of awareness included

medical/scientific journals and media sources such as television, newspaper, and the Internet. Interestingly, awareness was directly correlated with age; those older than 50 years of age were twice as likely to be aware as compared to those under 41. Of those who were aware, less than 20% had experience with a patient asking about DTC testing. Only five physicians had a patient bring in results from DTC genetic testing, and only one physician changed the patient's care based on those results. No physicians made a referral to a specialist such as a genetic counselor. Less than half of those aware thought DTC genetic testing was clinically useful; however, Family physicians were more likely to feel this testing was clinically useful as compared to Internal Medicine physicians. Additionally, 85% felt unprepared to answer a patient's questions regarding DTC genetic testing, and 71.5% wanted to learn more about it.

There are a few key points we can take from this study:

- In our small sample, physicians seemed unlikely to change patient care based on a patient's DTC genetic testing results. Four of five physicians did not alter their patient's care based on results.
- According to the survey, primary care physicians want to learn more about what DTC personal genomic testing involves, as well as obtain guidelines for management of patients who undergo such testing. Our study indicated that a lack of guidelines was a reason that physicians did not find testing clinically useful, and was why four of five physicians did not change patient care based on DTC results.
- As genetic counselors, we can help physicians understand how DTC genetic testing relates to patient care and how to discuss results with their patients when necessary. We can play a large role in educating physicians on new technology.

After graduation, I took a job at the Medical Center of Central Georgia. As the first genetic counselor employed by the hospital, I have been meeting with various physicians in the area to discuss the value of genetic counseling. Just the other day, I met with a Family Practice physician and the conversation turned to DTC personal genome testing rather quickly. I found myself thinking about my graduate school project and being surprised that I had stumbled upon one of the minority of Family Practice physicians aware of DTC genetic testing. Her colleague, another Family Practice physician, asked us what exactly this type of testing was. I spent time educating them and correcting some misunderstandings. I believe this experience reinforces the need for educating our primary care physicians about DTC genetic testing and human genetics in general. I hope that all genetic counselors will do their part in sharing DTC knowledge with our physician colleagues.

A special thanks to **Karen Powell**, **Carol Christianson**, and **Sonja Eubanks**, who were all part of my capstone committee. Thank you also to The UNCG Graduate Student Association and the Genomedical Connection who funded this project in addition to the Public Health Special Interest Group.

References

[1]Gurwitz D and Bregman-Eschet Y. Personal genomics services: Whose genomes? *European Journal of Human Genetics.* 17(7):883-9. 2009.
[2]Hogarth S, Javitt G, Melzer D. The current landscape for direct-to-consumer genetic testing: Legal, ethical, and policy issues. *Annual Review of Genomics and Human Genetics.* 9:161-82. 2008.
[3]Wasson K, Cook ED, Helzlsouer K. Direct-to-consumer online genetic testing and the four principles: An analysis of the ethical issues. *Ethics & Medicine: A Christian Perspective on Issues in Bioethics*. 22(2):83-91. 2006.
[4]Ohata T, Tsuchiya A, Watanabe M, Sumida T, Takada F. Physicians' opinion for 'new' genetic testing in Japan. *Journal of Human Genetics*. 54(4):203-8. 2009.
[5]Kolor K, Liu T, St Pierre J, Khoury MJ. Health care provider and consumer awareness, perceptions, and use of direct-to-consumer personal genomic tests, United States, 2008 [Letter to the editor]. *Genetics in Medicine*. 11(8):595. 2009.

NSGC News

The Collaborative Genetic Services Summit

By Angela Trepanier, MS, CGC

The NSGC's vision is to integrate genetics and genomics to improve the health of all. To this end, we are hosting **The Collaborative Genetic Services Summit** in mid-to-late summer 2011 using funds raised by the NSGC's Genetic Counseling Foundation (GCF).*

The purpose of the first phase of **The Collaborative Genetic Services Summit** is to bring together primary care providers, genetics specialists, and other key stakeholders to develop a skills and knowledge and, when available, an evidence-based collaborative model for integrating genetics and genomics into health care. The primary outcome of the Summit will be a consensus model(s) for triaging genetics and genomic services, which can be applied practically in the clinical setting to increase appropriate access to these services. A potential secondary outcome is that important gaps in evidence and in the genetics competencies of various health care providers needed to support the model will be identified.

Genomic Indication Triage Models

The project will involve developing draft triage models for five genomic indications that are representative of the majority of services that are currently offered, or will be offered in the future, through full genome sequencing. These include:

1. High-throughput population-based carrier screening
2. Cancer genetic risk assessment and testing using Lynch syndrome tumor tissue screening and testing as a model
3. Pharmacogenomic testing

4. Diagnosis of a congenital syndrome through full genome sequencing in the newborn period
5. Genomic profiling

Work Groups comprised of genetic counselors, physician assistants, nurses, clinical geneticists, primary care physicians, and other relevant health care providers will develop the draft models with the input and direction of the Project Investigators and an Advisory Committee. Physician Assistants and Nurses were chosen specifically because each group has already developed competencies related to genetics/genomics.

The first step in developing the models will be to delineate all the components of genetics services/counseling for each indication, from intake through long-term follow-up. The next step will be to highlight the practice-based competencies of different health care providers in delivering the genetics services/counseling required for each indication. This will involve looking at each profession's existing genomics competencies and evolving competencies (those being integrated into the training programs or in Continuing Education programs), and mapping them to the genetics tasks identified. Any available evidence from the biomedical literature will be incorporated at this step. The third step will be to develop the draft triage model(s) for genetic services/counseling based on indications for genetic counseling, case complexity, the knowledge and skills of various providers, and the available evidence.

The triage models are intended to be competency-based guidelines (rather than practice recommendations), by which an individual provider can self-assess whether s/he has the competencies needed to perform the roles associated with genetic services/counseling for a particular indication. The triage model(s) could also serve as a guide for payers to determine when consultant versus primary care services might be indicated.

Plans for the Summit

The draft models developed by the Work Groups and reviewed by the Investigators and Advisory Committee will then be vetted by a larger audience of stakeholders. This will occur during a one-and-a-half day Collaborative Genetic Services Summit, planned and hosted by the NSGC in Washington, D.C. Representatives from various health care professional organizations and other key stakeholder groups, including industry and advocacy organizations, will be invited to take part.

During the first day, an overview of the Summit goals and key issues, including relevant evidence, will be presented to the entire group to open the conference. Following this, participants will break into multidisciplinary groups to review the draft triage models. Attendees will discuss whether the models capture the essential components of genetic counseling, how important it is for their profession to be involved in these types of cases, whether there are barriers to their involvement, to what extent they are currently involved, and to what extent practitioners have the skills and knowledge to be involved. At the conclusion of the first day, the Investigators will collect the data from each group's discussion and make any needed changes to the draft triage models.

On the second day, the full group will convene for a facilitated discussion of the proposed models. Included in this discussion will be assumptions regarding the ability to

implement the triage models within the current health care system, possible limitations of each model, and additional evidence, education and/or resources necessary to support each model.

After the Summit, the Investigators will develop a draft white paper with the proposed triage model(s). These will be submitted to key professional and advocacy organizations for review. Further modifications may be made based on feedback, and a manuscript that delineates a model triage plan will be submitted for publication and distribution in 2012. The Investigators, with input from the Advisory Committee, will then begin to develop plans for the second phase of the Summit, which will address identified barriers to implementing the model triage plan.

The NSGC is Poised to Lead

We hope that the Summit and the work that precedes it will contribute in a meaningful way to discussions about how genetics services can be triaged effectively to promote access. We believe that this is a critical time to develop practical, competency-based triage models, and that by working in collaboration with a variety of health care professionals we can reach consensus. We also believe that in light of its mission, the NSGC is ideally suited and positioned to take the lead in this endeavor.

If you have any questions or suggestions, please contact **Angela Trepanier** at atrepani@med.wayne.edu.

**The GCF has been placed in a dormant state in 2011 and beyond, meaning that all active fundraising will halt. However, the Audrey Heimler Special Projects Fund and George Tiller Memorial Fund will remain under the GCF, so donors who wish to continue making tax-deductible donations can do so. In the event that an organization wishes to make a large donation toward the Summit or another future NSGC project that requires tax-deductible status, the GCF would still exist to accommodate the donation.*

Project Investigators

Angela Trepanier, MS, CGC (genetic counselor)
Joseph D. McInerney, MS, CGC (genetic counselor)
Michael Rackover, PA-C, MS (physician assistant)
Jean Jenkins, PhD, RN (nurse)
Frederick Chen, MD (Family Medicine physician)
Clinical geneticist (to be determined)

* * *

NSGC News

Call for Abstracts for the Audrey Heimler Special Projects Award

By Kristen Sund, PhD, MS

The deadline for the 2011 Audrey Heimler Special Projects Award (AHSPA) is **May 13, 2011**, so start thinking about your proposals today! Awards up to $5,000 are available to support projects that focus on the future of genetic counseling or the provision of genetic counseling services.

Some types of projects that might be appropriate for the AHSPA include:

- A pilot study to collect preliminary data for a larger future project
- Development of patient education materials
- Creation of tools for genetic counselors
- Development of novel ways to encourage leadership among new counselors

Additional details are available in the Members' Area of the NSGC website. Select the "Funding Opportunities" link for details about the application process. All proposals must be submitted to the Executive Office (nsgc@nsgc.org) by **May 13, 2011** for consideration. If you have questions about a proposal, please contact the Chair of the 2011 Audrey Heimler Special Projects Award Committee, **Kristen Sund, PhD, MS** at Kristen.Sund@cchmc.org.

ABGC Update

By the ABGC Board of Directors

The Board of Directors approved a number of new initiatives for 2011 and 2012 that are critical for the continued growth of the American Board of Genetic Counseling (ABGC) and the genetic counseling profession. We encourage all genetic counselors and Diplomates to become involved, even in small ways, to ensure the success of these initiatives. Ways to be involved include responding to survey requests and providing review and input as requested.

New Practice Analysis

The process to carry out a new Practice Analysis has begun! The Practice Analysis Advisory Committee (PAAC) met for their first time in January 2011. As you may recall, the purposes of a Practice Analysis are to determine the appropriate content matter for the credentialing examination, and to help define content areas that training programs seek to provide. (For more information on the first Genetic Counseling practice analysis process, see the Journal of Genetic Counseling article – Genetic Counseling Practice Analysis. The Practice Analysis survey is extremely important for the ABGC and the profession because the more data the PAAC has to analyze and consider, the more inclusive the examination content. Please respond to this survey and encourage all of your colleagues to respond as well. The survey should be distributed in **March 2011**.

Review of the Required Criteria for graduate programs in genetic counseling

The Accreditation Committee is undertaking a review of the Required Criteria, the governing document on standards for training programs. The purpose of this review is to improve the readability of the document for training programs wishing to be accredited and to update the components for best practices in accreditation as needed. This work will occur throughout 2011, and we anticipate completion in the first quarter of 2012.

Review of the Practice Based Competencies

The Board has also decided to embark on a review of the Practice Based Competencies. We plan to invite key stakeholders and representatives to participate in a strategic review of this document at a one- to two-day retreat in the fall of this year. The outcome of the retreat will be a full report on any necessary revisions to ensure that the Practice Based Competencies are current and reflect today's professional standards.

Transition Taskforce to begin separating accreditation and credentialing services

As announced at our Annual Business Meeting, the Board has begun the work of separating the accreditation and credentialing services into two distinct business agencies. This separation will accomplish a number of goals for the ABGC. First, this separation will eliminate the conflict of interest that currently exists between accrediting training programs and the requirements for entrance to the examination (graduation from an accrediting training program). Because of the growth and stability of the profession now, the ABGC is able to consider making this change while ensuring the survival of both new entities (credentialing genetic counselors and accreditation of training programs). The separation process will entail a great deal of planning and preparation. We anticipate that a complete separation will occur no sooner than 2013.

Each of these initiatives is essential for our profession's continued evolution and growth. The ABGC and each of these Committees and Task Forces will need the input of *all* genetic counselors – from the seasoned professional to the newly certified counselor. We thank those of you that replied to our request for volunteers, and we strongly encourage everyone to provide us with your input and insights.

Student Forum

Health Care Reform: President Obama's visit to Arcadia University

By Rebecca Mueller, BA, Arcadia University Genetic Counseling Program

March 7, 2010 was a Sunday spent like many others for students at Arcadia University's genetic counseling program –- we were studying. This particular day, we were cramming our brains with information for a midterm on cancer genetics: from a woman's lifetime risk of breast cancer, to *BRCA* mutation carrier rates and other facts on hereditary cancer. Memorizing and comprehending facts like these are not only critical for passing our course, but for counseling our patients accurately. We studied intently to ensure that the facts would stick.

Come Monday morning, however, these facts were eclipsed by a different set of facts. On March 8, 2010, President Obama came to Arcadia University to gain the support of Pennsylvanians for the Patient Protection and Affordable Care Act (PPACA), which was signed into law on March 23. An audience of nearly 2,000 people crowded into the Kuch Gymnasium, including a handful of students from the genetic counseling, physician assistant, and physical therapy programs, and faculty members. We collectively represented the health care field.

That morning, President Obama stood within twenty feet of us, describing the state of the health care system that we have inherited. He told the story of Natoma Canfield, a self-employed cancer survivor whose insurance charged her over $6,000 in premiums, and ultimately paid only about $900 for her care. He told the story of Laura Klitza, a young mother who is affected not only by the breast cancer that has spread to her bones, but also the tens of thousands of dollars of debt her medical bills have created – despite the fact that she has health insurance.

Third row back, starting third from left: Rebecca Mueller, Moriah Eberhard

People often critique the term "health care," arguing that the American system is really a "sick care" system, one that is skewed towards treating acute illness and away from ongoing management and preventive measures. As a genetic counseling student, I often think about the American health care system, and I stumble just as much over the word "care" as the word "health," contemplating whether patients typically receive consistent and comprehensive support. I question whether medical consultations, diagnostic studies, medical devices, and prescription medications are accessible to everyone, when so many are under- or uninsured.

Our Program Director, **Kathleen Valverde,** often draws our attention to the connotations of words; she says, "I know it is just semantics, but it matters." One distinction she draws is between "seeing" and "caring," encouraging us to ask ourselves, "Did I just *see* that patient or did I *care* for them?" In my opinion, a health care system largely based on exorbitant premiums and unaffordable co-pays, while denying coverage to those most in need, does not truly provide "care." In my vocabulary, "caring," means considering the medical and economic burden that illness so often imposes, in order to gather and disseminate information that aids clients in accessing available resources. It also means listening: to the father who declines a better-paying job because it would mean losing Medicaid coverage for his son with muscular dystrophy; to the young man with hemophilia whose company wants to let him go because his medical care is costing too much; to the recent college graduate with Gaucher disease whose new health insurance plan has medication co-pays that exceed her cost of rent. It means knowing whether Social Security stipulates coverage or support for people with conditions that we treat, and helping clients take advantage of pharmaceutical support plans. Once we understand how illness creates economic burden and how health insurance accessibility impacts life decisions, we can learn how Federal and State legislation may contribute to patient care.

The PPACA aims to help relieve some of the burden of health care costs by increasing access to affordable health insurance. It outlaws pre-existing medical condition clauses and makes attempts to help seniors with the potentially crushing cost of prescription drugs. The legislation is expected to be implemented over the course of the next eight years, if things go as planned. Political objection to the bill is substantial, threatening its full enactment in the months and years to come. In spite of the controversy, several changes have already been made over the course of summer and fall 2010. For example, insurers can no longer deny coverage to children with pre-existing conditions or impose lifetime limits on benefits. Furthermore, insurance companies must allow young adults to stay on their parents' health insurance plan until age twenty-six. For young people with chronic conditions or parents of children whose conditions interfere with coverage, this legislation could offer huge relief – if the families can afford the coverage.

Other measures outlined in the bill include abolishment of the above-mentioned pre-existing condition clauses to individuals of all ages, creation of "insurance exchanges" (state-brokered, regulated, standardized health care plans for eligible individuals and families), subsidies for people who are lower-income but above the poverty level, rebates for prescription drugs, and extension of Medicaid coverage. This legislation is important to those of us who counsel individuals and families struggling with health care expenses, because it may provide more affordable coverage to current and potential clients.

There are also some potential negative consequences associated with the PPACA. Many question the cost of the bill and the constitutionality of mandating individual coverage, arguing that it gives the government too much control over the health care system. The bill requires abortions be financed with private funds instead of federal funds, no doubt potentially putting many women in a difficult situation. The legislation also denies illegal immigrants access to state exchange plans, leaving that population uncovered. As members of the health care profession, these are issues that genetic counselors will likely be addressing frequently.

Proposed financing of health care reform will involve taxing higher income individuals and insurers of employer-based, high-cost health insurance plans, levying additional fees and taxes on health insurers, drug and device manufacturers and importers, and cutting Medicare spending. Those who fail to purchase coverage will be fined, including those individuals and large employers (greater than fifty employees) whose workers receive federal subsidies to gain coverage. The legislation also aims to control costs through a variety of measures, including the establishment of an Independent Medicare Advisory Board charged with cutting Medicare costs, funding for comparative effectiveness research, and the introduction of greater competition and transparency to the health care market. As genetic counselors are working to become Medicare providers, if enacted, the legislation will impact our profession and affect our clients (and our family and friends) by increasing health insurance options and potentially decreasing illness-related expenses.

Since its passage, the legislation has been widely challenged, with nearly two dozen federal lawsuits filed to block portions of the bill and a vote by the House of Representatives to repeal the bill altogether. Such challenges indicate that implementation of the legislation will be a complicated, contested, and costly process with an outcome that is uncertain at present. In March 2010, after noting that some felt

the reform was “too politically hard,” President Obama acknowledged “it is hard” because “health care is complicated.” No matter how politically difficult it may be to achieve, President Obama stated that this reform is “the right thing to do.”

In the coming months and years as the legislation is simultaneously implemented and challenged, the fight for reform is far from over. As constituents and genetic counselors, we should be politically cognizant of (if not active regarding) revision and enactment of the health care reform legislation. In doing so, we can explore potential ramifications for providers, and more importantly, we can start to understand the health care climate that dictates all of our lives. We can learn how the legislation may help (or burden) our clients and share this information with others who may be in similar situations. In this way, we can truly care for our clients, and begin to ensure that the health care system starts to live up to its name.

References

“Health care reform: Overview.” *New York Times online*. http://topics.nytimes.com/top/news/health/diseasesconditionsandhealthtopics/health_insurance_and_managed_care/health_care_reform/index.html?scp=1-spot&sq=health%20care%20reform&st=cse. Accessed on February 3, 2011.

Sahadi J. “Health reform; The $$$ story.” *CNNmoney.com*. Retrieved on March 25, 2010 from http://money.cnn.com/2010/03/20/news/economy/cbo_reconciliation/index.htm. Accessed on March 25, 2010.

The New Graduate Life

Three years in, and I’m still here

By Janelle McCarthy, MS, CGC

What is life like for recent graduates? I’ve been counseling for almost three years now and I still remember my very first day as a “genetic counselor.” I didn’t have a

supervisor, there was no one to report to after I took the family history, assessed the patient's risk, and offered testing. Instead, the patient was looking to *me* for answers. I was slightly beyond nervous, yet very excited. Looking back I think my first day went pretty well, considering the emotional rollercoaster going on privately in my own head that day, and during the days that followed.

I'll never forget the first time a patient asked, "And what does this have to do with *my* baby?" I thought, "Holy cow! Didn't I just spend the last fifteen minutes explaining that to you?"

The first time I had to give a patient bad news was memorable for a few reasons. Her pregnancy was affected with Down syndrome. I got all the information I could find: a list of parental support groups, a packet describing anticipatory guidance in care for different stages of life, and the options for termination at our facility. I thought about how I would react and what I would say when, not if, she started to cry. After all of that preparation I was amazed – there were no tears, but resigned acceptance, as she took the information and departed. As a new counselor, I was shocked at her unexpected reaction. Along with the shock was the surprise and relief I experienced because she didn't react in an emotional manner. Did this make me a bad counselor? Did I miss some body language or other non-verbal clue that I could have used to draw out the emotional reaction, which I'm sure she must have been feeling? Or was I just over-analyzing all of this?

My colleagues had their own first experiences as well, and we leaned on each other for support. One talked about her first patient to terminate a pregnancy and her experience sitting with the patient's family as they cried in the hallway when the patient started the procedure. Another coworker was newly experiencing the conflict and deep struggle over a disagreement she was having about the treatment of a patient by the physician and another genetic counselor. A different colleague verbalized the realization that the specialized job she took may not be exactly what she was looking to do, and she began questioning whether she had made the right choice. As I experienced being a new genetic counselor, I realized others were also sorting through similar issues and emotions.

The new experiences we have as recent graduates are not confined to work, but are also found in our personal lives with friends, family, and significant others. Most recent genetic counseling graduates are young and navigating through the life of a new working professional, both at the workplace and socially. Outside of our jobs, we may be dealing with other new events like planning weddings, having our first pregnancy (and integrating into it all our prenatal genetic counseling knowledge), acquiring the new role of "counselor" with family and friends as empathy skills overflow into personal lives, or searching for that "special someone" by going out on dates, partying and socializing, as often one's energy level permits. All the while, we are often encountered with the question, "And what do you do?" Sometimes we dodge the question and other times we embrace it, with the hope of bringing more light to our small field, one conversation at a time.

The balancing act required by new professionals is one that can be seen throughout the professional spectrum. The experiences and challenges we face as genetic counselors include the fact that, as much as our patients may affect us, we also have an impact on them. That is to say, how we feel and what we say can affect other people's lives on a

very personal level. The day that we are in a "bad mood" could be reflected in our words and interactions with other people, which could influence a specific action or a decision they make. It is a very humbling thought to know that we have so much influence on other people. However, it is also what is so rewarding about our profession. We have a subtle but palpable effect on others. That's one of the reasons I chose to be a genetic counselor; I wanted to be able to be in a profession that helps people. And with all the tribulations that come with being a young professional, I am comforted by the realization that what I do matters.

Nearly a month into being a genetic counselor, my mother asked me how was I liking my job. I answered, "It's challenging, but each day that I go home, I know that I have helped at least *one* person." Three years later, with all of the ups and downs of my private emotional rollercoaster, I have the satisfaction of knowing that if were asked the same question today, I would give the same answer. How many other young professionals can say the same thing?

Genetic Counselor Publications

By Jamie Fong, MS, CGC

Featured Article

By Denise Lautenbach, MS

Erby LA, Roter DL, **Biesecker BB**. Examination of standardized patient performance: accuracy and consistency of six standardized patients over time. *Patient Educ Couns*. Nov 19. 2010. [Epublication ahead of print]

There is no doubt that communication is an integral part of genetic counseling. But, after meeting with a patient, do you ever think about how you communicated information and whether or not that was the best way to go about doing so? Do you ever wonder why you presented information in a certain way, and whether or not there was evidence to support the effectiveness of the method you just employed? **Lori Erby, PhD, ScM, CGC** has recognized a lack of an evidence base for how we practice as genetic counselors. She has devoted much of her research efforts to thinking about ways to assess communication in genetic counseling, and how we can improve and lead to better patient outcomes.

Like many other research-based genetic counselors, Lori entered the Genetic Counseling Training Program at Johns Hopkins University (JHU) with the intention of becoming a clinical genetic counselor, having no idea that she would be interested in research at all. Then, as she worked on her Master's thesis project under the supervision of the highly dedicated and enthusiastic patient communication researcher, Dr. Debra Roter, Lori caught the "research bug." After finishing her Master's degree, Lori stayed at JHU to work on her PhD in Public Health, with a focus on social and behavioral research.

During her doctoral training, Lori was involved in the "Genetic Counseling Video Project" from its inception. The "Genetic Counseling Video Project" was a cross-sectional study of genetic counseling where genetic counselors were studied as they conducted simulated genetic counseling sessions with standardized patients. In this study, the standardized patients were graduate students or their friends with no previous training in genetic counseling or acting; Lori and the research team trained these individuals to act as patients for this study and follow a particular script, affect and communication style.[1] Lori helped to write the grant for this project and was the Project Director for this study during her doctoral training.

Since defending her dissertation in 2005, Lori now holds a faculty position at JHU, where she teaches, does research, and sees patients one day per week. Lori enjoys seeing patients in the clinic and also recognizes that keeping her "feet wet" in the clinical arena is critical to being a communication researcher. Actually, her clinical interaction serves as an important source of research questions as she reflects upon how she communicated and interacted with patients during each genetic counseling session.

While Lori participates in a number of research initiatives, her work with standardized patients in the context of genetic counseling offers the field of genetic counseling a great opportunity to study what we do. As our field is relatively small and we often have rare, but important, genetic counseling interactions, a study of communication using tapes of real genetic counseling sessions is not always feasible. Additionally, the use of standardized patients allows the researcher to control for variables such as race or age. Lori says that standardized patients are not only a research tool, but are currently widely used as a clinical training tool as well, providing the opportunity for students or various professional board examinees to be evaluated on clinical communication and interaction skills.

Lori encourages other genetic counselors to focus on increased involvement in genetic counseling research, even if just in small ways. Asking questions about *how* and *why* we do what we do, and how our methods and techniques may improve patient outcomes, will make a difference in our practice. Students are particularly poised to begin thinking about how research can be integrated early in one's career. After all, Lori's example shows that it is possible to jump right into research immediately after finishing a genetic counseling training program!

References

1. Roter D, Ellington L, Erby LH, Larson S, Dudley W. The genetic counseling video project (GCVP): Models of practice. *Am J Med Genet C Semin Med Genet.* Nov 15;142:209–20. 2006.

Articles co-authored by genetic counselors from June 2010 – January 2011

(Names of genetic counselors appear in bold)

Bachmann-Gagescu R, Mefford HC, Cowan C, Glew GM, Hing AV, Wallace S, Bader PI, Hamati A, Reitnauer PJ, Smith R, Stockton DW, Muhle H, Helbig I, Eichler EE,

Ballif BC, **Rosenfeld J**, Tsuchiya KD. Recurrent 200-kb deletions of 16p11.2 that include the *SH2B1* gene are associated with developmental delay and obesity. *Genet Med*. 12(10):641-647. 2010.

Brémond-Gignac D, Bitoun P, **Reis L**, Copin H, Murray JC, and Semina EV. Identification of dominant *FOXE3* and *PAX6* mutations in patients with congenital cataract and aniridia. *Mol Vis*. 16:1705-11. 2010.

Caleshu C, Day S, Rehm HL, **Baxter S**. Use and interpretation of genetic tests in cardiovascular genetics. *Heart.* 96(20):1669-1675. 2010.

Caleshu C, Shiloh S, Price C, Sapp J, **Biesecker B**. Invasive prenatal testing decisions in pregnancy after infertility. *Prenatal Diag*. 30(6):575-81. 2010.

Cohen, SA, **McIlvried, DE**. Impact of computer-assisted data collection, evaluation and management on the cancer genetic counselors time providing patient care. *Fam Cancer*. [Epublication ahead of print]. January 2011.

Erby LA, Roter DL, **Biesecker BB**. Examination of standardized patient performance: accuracy and consistency of six standardized patients over time. *Patient Educ Couns*. [Epublication ahead of print]. November 2010.

Faucett WA. International Standard Cytogenomic Array Consortium. Interview by **Alyson Krokosky**, Sharon Terry. *Genet Test Mol Biomarkers*. 14(5):585. 2010.

Gallego CJ, Grant J, Mikhail FM, **Barger C**, Robin NH. Use of array comparative genome hybridization in orofacial clefting. *J Craniofac Surg*. 21(5):1591-4. 2010.

Hadley DW, Ashida S, Jenkins JF, Calzone KA, Kirsch IR, Koehly LM. Colonoscopy use following mutation detection in Lynch syndrome: exploring a role for cancer screening in adaptation. *Clin Genet*. [Epublication ahead of print]. December 2010.

Hadley DW, Ashida S, Jenkins JF, Mensz JM, Calzone KA, Kuhn NR, McBride CM, Kirsch IR, Koehly LM. Generation after generation: exploring the psychological impact of providing genetic services through a cascading approach. *Genet. Med*. 12(12): 808-15. 2010.

Hock KT, Christensen KD, **Yashar BM**, Roberts JS, Gollust SE, **Uhlmann WR**. Direct-to-consumer genetic testing: an assessment of genetic counselors' knowledge and beliefs. [Epublication ahead of print]. January 2011.

Jablonski KA, McAteer JB, de Bakker PI, Franks PW, **Pollin TI**, Hanson RL, Saxena R, Fowler S, Shuldiner AR, Knowler WC, Altshuler D, Florez JC, Diabetes Prevention Program Research Group. Common variants in 40 genes assessed for diabetes incidence and response to metformin and lifestyle intervention in the diabetes prevention program. *Diabetes*. 59(10):2672-81. 2010.

McCuaig JM, Greenwood C, **Shuman C**, Chitayat D, Murphy KJ, Rosen B, **Armel SR**. Breast and ovarian cancer: Y do we forget about dad? *Lancet Oncol*. 11(12):1115-7. 2010.

McDonald-McGinn DM, Feret H, Bartlett SP, Whitaker LA, Zackai E. Metopic craniosynostosis due to mutations in *GLI3*: a novel association. *Am J Med Genet A*. 152A(7):1654-60. 2010.

Nowaczyk MJM, **Huggins MJ**, Fleming A, Mohide PT. Femoral facial syndrome: prenatal diagnosis and clinical features. Report of three cases. *Am J Med Genet A*. 152A(8):2029-33. 2010.

Powell KP, **Hasegawa L**, **McWalter K**. Expanding roles: a survey of public health genetic counselors. *J Genet Counsel*. 19(6):593-605. 2010.

Rosenfeld JA, Ballif BC, Torchia BS, Sahoo T, Ravnan JB, Schultz R, Lamb A, Bejjani BA, Shaffer LG. Copy number variations associated with autism spectrum disorders contribute to a spectrum of neurodevelopmental disorders. *Genet Med*. 12(11):694-702. 2010.

Rosenfeld JA, Crolla JA, Tomkins S, Bader P, Morrow B, Gorski J, **Troxell R**, Forster-Gibson C, Cilliers D, Hislop RG, Lamb A, Torchia B, Ballif BC, Shaffer LG. Refinement of causative genes in monosomy 1p36 through clinical and molecular cytogenetic characterization of small interstitial deletions. *Am J Med Genet A*. 152A(8):1951-9. 2010.

Rosenfeld JA, Lacassie Y, **El-Khechen D**, Escobar LF, Reggin J, **Heuer C**, Chen E, Jenkins LS, Collins AT, Zinner S, Babcock M, Morrow B, Schultz RA, Torchia BS, Ballif BC, Tsuchiya KD, Shaffer LG. New cases and refinement of the critical region in the 1q41q42 microdeletion syndrome. *Eur J Med Genet*. [Epublication ahead of print]. October 2010.

Rutlege KD, Barger C, Grant JH, Robin NH. *IRF6* mutations in mixed isolated familial clefting. *Am J Med Genet A*. 152A(12):3107-9. 2010.

Schimke RN, **Collins DL**, Stolle CA. Paraganglioma, neuroblastoma and a *SDHB* mutation: resolution of a 30 year old mystery. *Am J Med Genet A*. 152A(6):1531-5. 2010.

Shen H, Damcott CM, Rampersaud E, **Pollin TI**, Horenstein RB, McArdle PF, Peyser PA, Bielak LF, Post WS, Chang YP, Ryan KA, Miller M, Rumberger JA, Sheedy PF 2nd, Shelton J, O'Connell JR, Shuldiner AR, Mitchell BD. Familial defective apolipoprotein B-100 and increased low-density lipoprotein cholesterol and coronary artery calcification in the Old Order Amish. *Arch Intern Med*. 170(20):1850-5. 2010.

Sung RK, Ursell PC, Rame JE, Bailey, H, **Caleshu C,** Nussbaum, RL, Scheinman, MM. QTc prolongation and family history of sudden death in a patient with desmin cardiomyopathy. *Pacing Clin Electrophysiol.* [Epublication ahead of print]. July 2010.

Sutphen R, Davila B, **Shappell H**, Holtje T, Vadaparampil S, Friedman S, Toscano M, Armstrong J. Real world experience with cancer genetic counseling via telephone. *Fam Cancer*. 9(4):681-9. 2010.

Teman E, Ivry T, **Bernhardt B**. Pregnancy as a proclamation of faith: Ultra-orthodox Jewish women navigating the uncertainty of pregnancy and prenatal diagnosis. *Am J Med Genet A*. 155A(1):69-80. 2011.

Terry SF, **Krokosky A.** Rare conditions: where do primary care and genetic diseases intersect? *J Am Assoc Physician Assistants*. 23(11):63-4. 2010.

Zemble R, Luning Prak E, McDonald K, **McDonald-McGinn D**, Zackai E, Sullivan K. Secondary immunologic consequences in chromosome 22q11.2 deletion syndrome (DiGeorge syndrome/velocardiofacial syndrome). *Clin Immunol*. 136(3):409-18. 2010.

Letter to the editor co-authored by genetic counselor

(Name of genetic counselor appears in bold)

Chiang P-W, **Thomas M**, Spector E, Frei-Jones M. Novel mutation causing Hermansky-Pudlak syndrome type 2. *Pediatr Blood Cancer*. 55(7):1438. 2010.

Book co-authored by genetic counselors

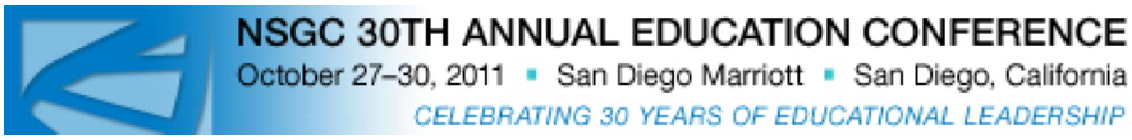
(Names of genetic counselors appear in bold)

Peay HL and **Austin JC**. How To Talk With Families About Genetics and Psychiatric Illness. WW Norton & Co. 2011.

Please send references of published articles by genetic counselors to Jamie Fong at jaf2025@med.cornell.edu

AEC Update

NSGC 30th Annual Education Conference



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

We are excited to invite you to join us in San Diego, California for the 30th Annual Education Conference (AEC). You will soon receive your 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 thirty years of educational leadership with you.

Plan Ahead: Stay for the Entire AEC

In response to the membership's desire to shorten the overall length of the AEC without cutting the number of CEU opportunities, the AEC debuted a new format for the 2009 meeting in Atlanta, Georgia. While we continue to receive ongoing feedback related to the schedule, dates for the 2011 conference were already locked in, so the format will remain the same for 2011. Please know that your feedback is being heard, and the Board is considering AEC format changes for future AECs.

The 2011 AEC will again begin with the "Welcome to the AEC" orientation, followed by the opening plenary Janus Series and Best Abstract Awards. Concluding this kickoff will be the Welcome Reception in the Exhibitor Suite on Thursday evening. There will be three full days of outstanding educational opportunities within the Plenary and Educational Breakout Sessions on Friday and Saturday, followed by a shorter day on Sunday and the conference's conclusion in the late afternoon. East coast attendees, in particular, may wish to fly home on Monday so that you are able to stay for the entire conference; you won't want to miss opportunities for learning and CEUs that occur later on Sunday.

Pre-Conference Symposia

Based on the positive feedback from the past two years, we will again have six Pre-Conference Symposia on the opening day, Thursday. The Pre-Conference Symposia are high level, in-depth sessions for specific specialty practice areas, new issues in genetics and genomics or professional development topics. Each session will last five hours, allowing for a deeper review and discussion of a particular topic. The attendance at each symposium will be smaller than at the Educational Breakout Sessions, which will allow

for a more interactive experience. Pre-Conference symposia will require separate registration from the AEC and will have limited space available. Sign up early!

Continuing Education Units

The NSGC is approved as an Authorized Provider for CEUs through the International Association for Continuing Education and Training (IACET). IACET requires that we document attendance for the sessions for which individuals are requesting CEUs. The 2011 AEC will again be using bar-code scanners to quickly scan attendee badges as attendees enter a session.

Program Book

In an effort to reduce costs and "go green," handouts will be available online prior to the conference, for attendees to print themselves. We recommend that you review the conference handouts prior to arriving in San Diego and print ones you want to have on paper during the conference. Another option, if you have a laptop, netbook or tablet computer, is to download handouts for viewing on your device during the presentations.

Accommodations

The AEC will be held at the waterfront San Diego Marriott Hotel and Marina, which is less than five miles from the San Diego International Airport (SAN). The Marriott provides beautiful views of the San Diego Marina and is adjacent to Seaport Village, which features ample waterfront dining and shopping. The Gaslamp Quarter, also within walking distance, is famous for its great restaurants and shopping. The world famous San Diego Zoo is less than five miles from the hotel, as is historic Old Town San Diego.

Dates to Remember

The deadline for early registration for the AEC is **August 31, 2011**. Be sure to sign up on time to take advantage of this discount! Abstracts for platform or poster presentations will be accepted from **March 21 to May 16, 2011**. See the NSGC Web site for more information.

The NSGC's 30th AEC promises something for everyone. Mark your calendars now to join us in San Diego, California!

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 Robin M. Troxell, MS, CGC

The Immortal Life of Henrietta Lacks
Author: Rebecca Skloot
Publisher: Crown, First Edition, 2010
Pages: 384
Retail price: $26.00
ISBN-10: 1400052173

Henrietta Lacks, an African-American woman from Clover, Virginia, lived only thirty years but cells from her malignant cervical tumor live on as HeLa in thousands of laboratories worldwide. The Immortal Life of Henrietta Lacks is a comprehensive look at the Lacks family and the turmoil brought on by the use of Henrietta's cells without consent, confidentiality, or compensation. Author Rebecca Skloot covered thousands of miles and several generations of the Lacks family to tell the most complete story to date of this family and the impact HeLa cells have had on science.

The book begins with Henrietta's initial biopsy at Johns Hopkins by Dr. Howard Jones, then takes the reader back in time to 1920 when Henrietta Lacks was born Loretta Pleasant. The author describes her childhood and how she grew up with her cousin and future husband, Day Lacks, farming tobacco with their grandfather and myriad other relatives. Their first child was born when Henrietta was just fourteen years old and they eventually married when she was twenty. By the time she was evaluated at Hopkins in 1951, her fifth child was three months old. Henrietta's initial biopsy results were (erroneously) classified as "epidermoid carcinoma of the cervix, Stage I" necessitating the need for treatment. At that second visit, prior to her first treatment with internal radium, two small samples were taken and sent to Dr. George Gey, the head of Tissue Culture Research at Hopkins. The sample from the cervical tumor did what no other sample had done before – survived in culture and, in fact, thrived. Named "HeLa," the cells were initially shared by Dr. Gey with any scientist who was interested, and eventually would be instrumental in not only cancer research, but also developing the vaccine for polio, atomic bomb testing, and genetic research.

About twenty years after her death, an article in *Obstetrics and Gynecology* written by Dr. Jones, Dr. Victor McKusick, and others in posthumous tribute to Dr. Gey's career, detailed the origins of the original HeLa cell line and used Henrietta's full name. That article, plus a chance encounter between a National Institutes of Health researcher and a member of the Lacks family, set in motion the Lacks family's discovery of their mother's "immortal" cells, and the quest of Henrietta's only living daughter, Deborah, to find out who her mother was, what happened at Hopkins, and who took her cells without consent.

Eventually, this also led to the author's decade-long journey of researching and writing this book. She raises many ethical, legal and social issues, including informed consent, use of minorities for research, funding for research, and who benefits from the use of tissue samples. Some family members were angry that samples were taken without consent, some were angry that her name was often misquoted as Helen Lane or Helen

Larson, and some were furious that HeLa helped launch a multi-billion dollar tissue culture industry while the majority of the Lacks family were so poor they could not afford health insurance and often went without treatment for diabetes, heart disease, and hypertension – ironically, all diseases for which HeLa cells helped benefit research and treatments. This book centers on Deborah's search to find out what happened to her mother, to her sister Elsie who died in an institution shortly after Henrietta's death, and the contributions HeLa cells have made to the world.

The Immortal Life of Henrietta Lacks is well written, meticulously researched, and presents events spanning more than eighty years in an organized fashion. Any individual working in the field of genetics or medical research or who has an interest in the history of ethical, legal and social challenges would benefit from reading this book.

Research Network

By Emily Place, MS, CGC

Brain, Behavior and Genetic Studies of 22q11.2 Deletion

The Division of Genetics at The Children's Hospital of Philadelphia (CHOP) and the Department of Psychiatry and Neuro-Imaging at University of Pennsylvania School of Medicine are teaming up to study the brain and behavior in patients with the 22q11.2 deletion. This five-year study, supported by the National Institute of Mental Health, is open to individuals over the age of 18 years with 22q11.2 deletion. The objectives are to combine genetic and neurobiologic paradigms for understanding pathogenesis, and for detection of genes that modulate susceptibility to psychosis with phenotypic features of schizophrenia and related disorders. Principal Investigators include basic scientist Beverly Emanuel, PhD at CHOP, and psychiatrist researcher Raquel Gur, MD at Hospital of the University of Pennsylvania, in collaboration with Donna McDonald-McGinn, MS, CGC, Program Director for the "22q and You" Clinic at CHOP and Elaine Zackai, MD, Medical Director at CHOP.

Contact: Study Coordinator, Margaret Sounders, MSN, PhD at 215-590-2920 or souders@email.chop.edu

Asthma Medications in Pregnancy Surveillance System (AMPSS)

The Organization of Teratology Information Specialists (OTIS) is researching the use of short-acting versus long-acting beta-agonists for the treatment of asthma in pregnancy. Pregnant women may qualify for participation if they have asthma and have been using an asthma medication during pregnancy. OTIS is also enrolling controls for this study (women who do not have asthma and have not used a medication for the treatment of asthma during their pregnancy).

Contact: (877) 311-8972 or http://www.otispregnancy.org/ongoing-research-studies-p135738

Vaccines and Medications in Pregnancy Surveillance System (VAMPSS)

The Organization of Teratology Information Specialists (OTIS) is researching vaccines and medications in pregnancy such as the *H1N1* vaccine, seasonal flu vaccine, and antiviral medications. Participants will not be asked to take any medication or vaccines as part of this study. Eligible participants will be pregnant women who have already received the vaccines or taken antiviral medications. Pregnant women who have not received the vaccines or antiviral medications are also eligible to participate as controls. Visit the website to learn more about this study.

Contact: (877) 311-8972 or http://www.otispregnancy.org/ongoing-research-studies-p135738

FaceBase Biorepository

The FaceBase Biorepository is recruiting individuals with craniofacial anomalies, especially cleft lip and palate, to serve as a resource for investigators studying these disorders. Individuals with both syndromic and non-syndromic craniofacial anomalies and their family members are eligible to participate. Participation involves providing a biological sample and completing a questionnaire. Referring clinicians are asked to send relevant medical records. Samples and data will be provided to researchers as de-identified; participants and the referring clinician(s) will receive general research updates.

Contact: Kate Durda MS, CGC at 1-866-520-8982 or 319-335-9632, kate-durda@uiowa.edu or Jeff Murray, MD at jeff-murray@uiowa.edu

Project FLAG

This multi-center, National Cancer Institute-funded study of hereditary gastrointestinal stromal tumors (GIST) is led by Judy E. Garber, MD, of the Dana-Farber Cancer Institute in collaboration with co-investigators at the Memorial Sloan-Kettering Cancer Center and MD Anderson Cancer Center. The study is open to individuals age 18 years and older with GIST. The study goals include defining the phenotype and genetic basis of inherited GIST susceptibility, as well as other benign or cancerous familial GIST associations. Participants meeting protocol-defined high-risk criteria will have the option of germline genetic analysis of genes *KIT* and *PDGFRA*.

Contact: Individuals can enroll at www.ProjectFLAG.org or by calling 1-800-828-6622, option #1.

Genetic Epidemiology of Lung Cancer Study

This is a study of increased genetic susceptibility to lung cancer. The highly experienced group, Genetic Epidemiology of Lung Cancer Consortium (GELCC), has found the first familial lung cancer gene *RGS17* (located on chromosome 6q23) and has linked a chromosome 15q variant (associated with nicotine dependence) to a subset of lung cancer families. The study is collecting lung cancer cases with a limited family history (two lung cancers on one side of the family) for genome-wide association studies and families with four or more cases of lung cancer, living or deceased, on one side of the family for linkage studies.

Contact: Debra O'Connell at (419) 383-4341 or debra.oconnell@utoledo.edu or Principal Investigator Colette Gaba at (419) 383-4557 or colette.gaba@utoledo.edu

Inherited Eye Disease

The University of Iowa, John and Marcia Carver Nonprofit Genetic Testing Laboratory is conducting research to identify genes, and the genetic variations within these genes, which cause inherited eye diseases and identify the clinical features associated with these genetic variations. One example of this work is Project 3000 (www.project3000.org). Project 3000 is a nationwide initiative to genotype every patient in the United States with Leber Congenital Amaurosis (LCA). As *RPE65-related* gene replacement therapy trials for LCA appear promising, a molecular diagnosis for LCA is increasingly relevant.

Contact: Tiffany Grider, MS, CGC at (319)353-7242 or carverlab@uiowa.edu**;** www.carverlab.org

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