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President's Beat

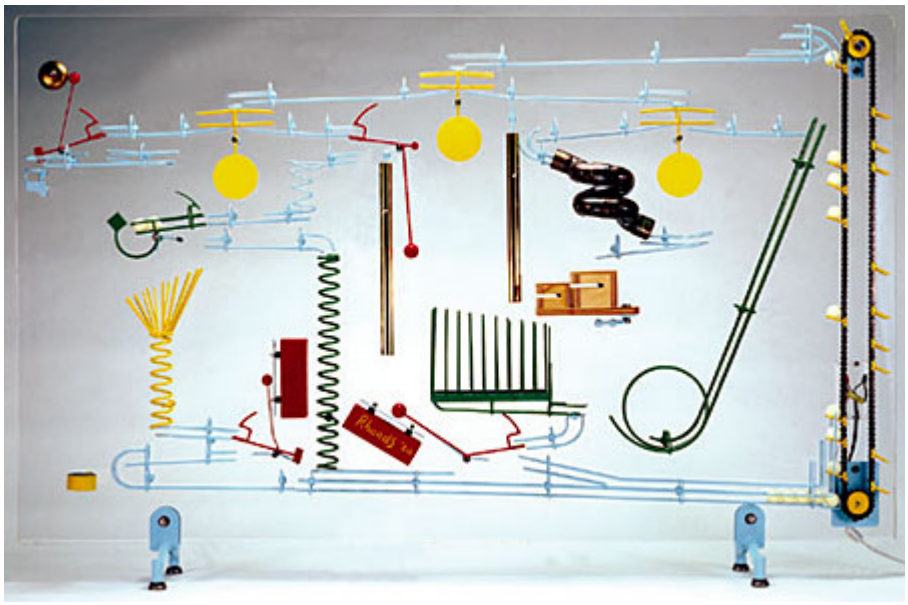
Small is to Big as Plates are to Golf Balls

Those of us who have been around since the early days of the National Society of Genetic Counselors (NSGC) fondly recall the collegiality of belonging to a small, select group of professionals. Even today, with over 2,700 NSGC members and more than 3,000 total practitioners, we are still a small profession. I can think of few occupations in which so many of us know each other on a first name basis, not to mention the added bonus of annual reunions with our graduate classmates at professional meetings. That being said, today's NSGC is a much different organization than it was even ten years ago. Taken together, the NSGC's various activities involve over 1,000 volunteers – a staggering testament to member engagement in our professional society! As President, I keep a flow chart of the society's governance structure and leadership on my desk for handy reference. In 2012, the NSGC is composed of:

- 8 full Committees
- 2 Subcommittees
- 2 Working Groups
- 19 Special Interest Groups
- A 12-person Board of Directors
- Countless individual contributors

While the bulk of the NSGC's work is accomplished through the efforts of our member volunteers, we benefit greatly from the expertise of our contracted management firm, SmithBucklin, through our Executive Director Meghan Carey, the four additional members of the administrative staff team, two government relations staff, and ten additional staff spending part of their time on marketing, accounting, Annual Education Conference logistics, and other functions for the NSGC. Given all the moving parts involved in the organization's work, I used to think of the NSGC as deftly spinning several plates for the benefit of our profession. I'll take a page from Kurt Vonnegut here and provide a picture to illustrate my point:

My bird's eye view as a Board member and now as President has made me realize that the plate spinning analogy is inadequate to describe the complex and layered network of activity that is today's NSGC. A more apt analogy nowadays would be one of those ubiquitous kinetic golf ball sculptures by George Rhoads that are found in public places from children's hospitals to museums. Here's one that especially reminds me of the NSGC's current governance structure:



I like the golf ball sculpture analogy because it illustrates a number of key aspects of the NSGC that may not be apparent on casual observation. Like the various components of the sculpture, the NSGC's diverse activities are intertwined and inter-dependent. A decision in one committee might have unintended ripple effects on other, seemingly unrelated work. Keeping track of the "big picture" is something that our Board of Directors and Executive Office focus on daily. Another aspect of a large golf ball

sculpture, and part of what makes it so interesting, is that many things happen at once. Unless you're a particularly astute and practiced observer of such structures (as I used to be when my children were small!), it's hard to follow everything that's happening. If you're focused on the ascending escalator of golf balls heading toward the loop-de-loop, you may not notice the spiraling queue of golf balls that topples the swinging bucket at the precise moment it reaches a critical mass. Hopefully, you see my point: in a growing and strategically ambitious organization, it's sometimes hard to discern all the interconnections that move us toward achieving our goals.

One final characteristic of a golf ball structure is that all of its inner workings are visible. If its surrounding glass enclosure weren't transparent, the final sculpture would be irrelevant, even if all the golf balls ultimately ended up at their final destinations. With such a large percentage of member engagement, it would be difficult to avoid transparency within the NSGC. Still, at each meeting of our Board of Directors, we invariably schedule time to talk about ways to improve communication with the membership and to emphasize the transparency of our policies and processes. The Board recently considered recommendations stemming from a comprehensive "communications audit" and will be prioritizing some exciting upgrades for 2013 – stay tuned! That being said, as the golf ball sculpture illustrates, not all of the inner workings of a complex kinetic structure will be readily apparent from all vantage points. Fortunately, genetic counselors are never shy about asking for clarification when something is unclear, as evidenced by the hundreds of questions and suggestions fielded by our Executive Office each year through email, telephone, and on our Discussion Forums. Rest assured that if you're thinking about a professional issue, chances are that the NSGC is also thinking about it, right there in the center of the maze where the rubber hammer hits the gong. Call us or email us if you can't see it, and we'll bring it to the forefront. We value your input, we thrive on new ideas, and we strive to serve as the voice for our enthusiastic and vibrant profession.

Brenda Finucane, MS, CGC
2012 NSGC President

The 2011 NSGC Cultural Competency Scholarship Award Recipients: Their Stories

Lauren Grote, MS

The 2010 United States Census Data reported the population as 72.4% White, 12.6% African American, 0.9% American Indian or Alaskan Native, 4.8% Asian, 0.2% Native Hawaiian or Other Pacific Islander, 6.2% of another race, and 2.9% two or more races; ethnicity is reported as 16.3% Hispanic or Latino.[1] It is clear that the number of minority individuals living and starting families in the U.S. is continually increasing. According to the Pew Research Center, by 2050 one in five Americans will be an immigrant and "Whites" are expected to become a minority, making up only 47% of the population.[2]

These numbers alone should serve as an incentive for the genetic counseling field to become more culturally competent. The National Society of Genetic Counselors (NSGC) conducts a Professional Status Survey every two years. According to the 2012 survey, respondents reported the following races: 92% White or Caucasian, 5% Asian, 1% Black or African American, 1% Other, 0.4% Native Hawaiian or Other Pacific Islander, and 0.1% American Indian or Alaskan Native. Genetic counselors will be increasingly counseling individuals of races, ethnicities, and cultures different than our own, and we need to be prepared to offer support to these individuals. We will begin to see people with and counsel more often about disorders that were once rarely seen because there are now more individuals of other ethnicities, races, and cultures emigrating from their home countries. As a genetic counselor, I have personally vowed to take all of the steps I can to help my patients feel at ease and have confidence in my role as part of their health team, regardless of their cultural backgrounds.

Prior to receiving one of the NSGC Cultural Competency Scholarships in 2011, I worked as part of a team with **Nancy Steinberg Warren** and many others to help design and write case studies for the Genetic Counseling Cultural and Linguistic Competence Toolkit, which is accessible at http://geneticcounselingtoolkit.com. The Toolkit contains case studies, teaching tools, and clinical tools, among other things, to help advance one's

knowledge of cultural competency in his or her field. My participation in this project was how I first became interested in intertwining cultural competency and genetic counseling. I began to formulate ideas about how I could become a culturally competent genetic counselor some day when I started my career. It was important for me to remember that someone's culture can be as simple as the way they think and the things they believe.

As I began counseling patients as a genetic counseling student, I found out that these things are not as simple as I expected. I learned that most of the time, you have limited information about a patient; even if you do know the patient's country of origin, preferred language, or ethnicity, you cannot predict what beliefs, preferences, or knowledge they may have based on this information.

I received the scholarship early in my second year as a genetic counseling student, and initially it helped fund my attendance at the NSGC's 2011 Annual Education Conference. However, more importantly, it has continually opened my eyes and my counseling to be sensitive to cultural competency. It helped me further establish my own interests, and also helped the interest of my classmates flourish when I completed presentations and assignments about cultural competency. As a close-knit class, we discussed cases that challenged us due to a cultural issue, and learned how to expand our knowledge from what we learned in class to what we experienced in clinic. Each encounter with a patient of another culture was a learning experience that helped not only ourselves, but also each other, as we shared both good and bad sessions with our peers.

Receiving this award encouraged me to expand my confidence in being a culturally competent genetic counselor. Towards the end of my second year in graduate school, I gladly took on opportunities to counsel individuals who needed an interpreter, came from another country to receive services at the hospital, or simply had a different ethnicity than I did. For some cases I was extremely prepared, while for others I found myself totally surprised and thoroughly unprepared.

I think these experiences taught me that becoming culturally competent is a continuous process. It is a task I will strive to excel at every day as a genetic counselor. I must also keep in mind that each person I counsel will likely have influences from a certain culture, experiences that have changed their viewpoints, and beliefs they hold due to the course their life has taken. Although these nuances cannot be predicted, I can learn to adjust to them during a session and help incorporate a patient's belief system into their plan of care. I will strive to be aware that every person is a distinct individual, and that I need to let go of any preconceived notions about his or her life. In doing this, I will be able to let the patient educate me on his or her belief system, preferences, and cultural experience, which I believe is the most important aspect of being a culturally competent genetic counselor.

Throughout this process, I learned how to evaluate myself as a culturally competent person both in the personal and professional worlds. I would encourage any student who is interested in cultural competency issues to apply for this scholarship. It has benefited

my career as a student and as a new genetic counselor. It is an excellent opportunity to further your knowledge and experience in the field of cultural competency.

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Seeing Through the Cultural Awareness Glasses

Julia Su, MS

In October 2011, I had the honor to stand at the podium of the National Society of Genetic Counselors' (NSGC) Annual Education Conference in San Diego to receive one of the NSGC Cultural Competency Scholarships. Through researching and composing the application essay, I have become much more aware of the issues in multiculturalism in the context of genetic counseling. The curiosity in me to learn more about different cultures was rekindled and reconfirmed through this process. This curiosity and interest continued long after receiving the scholarship. In my rotation as a second year genetic counseling student and in my work as a genetic counselor after graduation, I paid more attention to the cultural cues, both verbal and non-verbal. I began to draw connections I had never thought of before, and to ask questions that are outside of my typical "thought box." I was learning to see through "cultural awareness glasses."

I remembered that as a second year genetic counseling student at Sarah Lawrence College, I had the opportunity to co-counsel a family with a boy with Klinefelter syndrome. The family was of Indian descent. Because of the set-up of the appointments, the child did not come to the appointment, and the purpose of the counseling session was to talk with the parents. As I greeted the parents in the waiting room, I noticed the "red

dots" on their foreheads and the parents' contrasting outfits: the mother was in a colorful traditional Indian dress, while the father in a formal business suit. As we walked down the hallway to the counseling room together, I could hear Hindi music coming out of the portable music player the mother was holding. After my supervisor and I entered the counseling room and introduced ourselves, the parents greeted us with very polite bows, to which we responded with bows, instead of the usual handshakes we give.

During the counseling session, the father revealed to us that he works as a pediatrician, and he was actually the first person to suspect Klinefelter syndrome in his son. While the father did most of the talking, the mother was very quiet, gently nodding to her husband in agreement, and would only speak when my supervisor or I directed a question to her. In our meeting, we went on to discuss many aspects of Klinefelter syndrome, including both the medical management and the psychosocial implications. The family shared with us their high hope for fertility in their son and the wish to "continue the family line." We touched on sensitive topics like how they viewed this illness in the context of their religious practice of Hinduism. We also discussed the father's mixed feelings resulting from playing both roles of a dad and a doctor.

Throughout the session, I felt my supervisor and I were on a journey together with this couple, exploring with them what it meant to have a son with Klinefelter syndrome. Moreover, we were exploring what it meant to have a son with Klinefelter syndrome in this Indian family, who highly valued their faith and the prospect of having a large biological family. In addition, this was also a family where the father was walking the tight rope between a well-intentioned parent and a well-informed pediatrician, while the mother quietly upheld her standard of a "good wife," which could have been attributed to her cultural background.

After the session, I went on to do some research on my own. I read books and articles, and talked to other genetic counselors. I learned that the "red dot" that some Indian women wear on their foreheads is commonly called a *bindi*. I also read about what it means to wear one, and when and how a *bindi* might be worn. I gained a good general overview of Indian culture and Hinduism from books: the philosophy, festivals, and rituals, etc, and I was eager to learn more from other sources as well.

As well, my supervisor and I reflected together about the way we handled the case. We asked ourselves questions like, was it appropriate that we responded to the family's bows with bows instead of handshakes? Was it right to try to involve the mother in conversations through directing questions to her? Would the father think we were undermining his authority in the family by doing so? This case was only one of many I have experienced in clinic. Whenever I encountered cases with unique cultural issues, I often thought back to my experience with the Cultural Competency Scholarship and felt grateful that my participation in the scholarship had heightened my sensitivity and interests towards these issues.

The practice of putting on "cultural awareness glasses" didn't stop as I left my genetic counseling training program. After graduation, I accepted a position as a genetic

counselor in a regional newborn screening program, primarily responsible for retrieving positive newborn screening cases and helping families cope with the follow-up testing process. In this role, I see how families consciously and subconsciously make decisions under the influences of their cultural backgrounds, including decisions about birth locations, feeding choices, and whom to bring to the genetics appointment. In one counseling session, aside from the newborn screening topic we discussed, my patient, a young Chinese woman, and I chatted about her reluctance to supplement breastfeeding with formula, as advised by the pediatrician due to her baby's slow weight gain. She told me that as breast milk has always been deemed as the most pure and harmonious food for infants in the region her family came from in China, she felt torn by having to introduce her baby to formula, something that was thought of as "synthetic" and "of lower grade". Though certainly not the only factor, culture is an important one in shaping my patients' interpretations of the meaning of birth, caring for a newborn, and the expansion of a family.

However, during my work, I've also come to realize that a cultural observation is always retrospective. It is only after our patient has made a certain choice that we can we look back at the situation and reflect on the role culture has played in it. Culture, tradition, or anything else we think we know about our patients, doesn't give us the power to predict what our patients would choose or how they would react. Interestingly, this lack of ability to predict is exactly one of the key factors in making our job as genetic counselors so fascinating, as well as keeping us humble in our professional journey.

For the past nine months in my role as a second-year genetic counseling student and later as a new genetic counselor, I have continued to ponder the same question that initially motivated me to participate in the Cultural Competency Scholarship: What does cultural competency really mean in the context of genetic counseling? The more I looked, the more I realized how multi-faceted and multi-dimensional the answer to this question could be. I know my journey of exploration will continue, and I will be sure to keep my "cultural awareness glasses" on. As this Cultural Competency Scholarship helped me further my education, broadened my perspectives, and enriched my genetic counseling training experience, I would definitely encourage any second year genetic counseling student who is interested in multicultural issues to apply for the 2012 NSGC Cultural Competency Scholarship.

A Unique Opportunity to Broaden My Genetic Counseling Training Through a Nontraditional Experience

By Corrie Bourdon, Indiana University Genetic Counseling Program Class of 2013

When I got a phone call from Lineagen, Inc. (Lineagen) offering me the opportunity of a month-long rotation to learn about their unique company, I did not realize how much I could learn from such a compassionate and patient-focused team. My introduction to Lineagen began at the 4p- Support Group conference where genetic counselors from the company were able to meet with families affected by Wolf-Hirschhorn syndrome and related conditions.

Many of these families described their first interactions with genetics professionals as being very negative. They were told their child would not live very long, nor have a personality. After meeting with the Lineagen genetic counselors, who listened to their stories, answered questions about genetics, and explained the technology behind Lineagen's high-density chromosomal microarray (CMA) testing, these families expressed their gratitude for being able to share their experiences and their excitement to learn more about their childrens' genetics. The purpose of offering CMA to these families, whose children were originally diagnosed by fluorescence *in situ* hybridization (FISH) testing or a G-banded karyotype, was to identify the specific breakpoints of each child's deletion and to identify any unbalanced rearrangements. By discovering the specific breakpoints, clinical management of these children might change based upon the breakpoint locations and the genes impacted by the deletion.

My official rotation began with understanding how Lineagen's mission impacts every aspect of the way they run as a company. They aim to accelerate and enhance the diagnostic evaluation of individuals with medical conditions so the best possible outcomes can be achieved for patients and their families. It was their commitment to autism spectrum disorders (ASD) that first piqued my interest to find out more about this company.

Before entering my training program, I worked as a behavioral therapist and developed a passion for children and families affected by ASD. As a result of that experience, I have

personally witnessed the many obstacles families go through, extensively searching to find the reason behind their child's diagnosis of ASD. They want to understand the etiology and give their child the best care for their developmental needs. To help these families, it is important for physicians to have the necessary tools for efficient and appropriate genetic diagnoses, if they are warranted. Lineagen helps make that a possibility by educating community physicians about their CMA and fragile X testing (called FirstStep^{Dx}) and genetic counseling services. This process aims to accelerate the diagnostic evaluation so these patients and their families can receive the most appropriate services and future evaluations by medical geneticists and other physicians.

It has been enlightening to witness the ways Lineagen's entire team collaborates in a dynamic environment. They are continually evolving to maintain the highest level of patient care, while rising to meet demands of insurance agencies and reaching out to community physicians about the necessity of CMA testing for children with developmental delay (DD) or ASD. To do this, the Lineagen team strives to be transparent with one another within the company, the genetics community, and most importantly, the patients. They participate in company-wide weekly meetings to discuss achievements, setbacks, and goals and are actively involved with organizations in the genetics community. In addition, the Lineagen team strives to ensure that FirstStep^{Dx} is appropriate for a family based on its specific clinical and psychosocial situation, and communicates with families throughout the testing process regarding any out-of-pocket costs the family will have.

Lineagen is also unique in their use of alternate service delivery models. As a genetic counseling student, I have spent the majority of my training learning about and being immersed in clinical genetic counseling. Because of this focus, my experience with alternate service delivery models, specifically telephone counseling, has been limited to calling out results to patients and coordinating care between clinic visits. Lineagen's model gives families and physicians the opportunity for pre-test and post-test telephone genetic counseling, along with a personalized electronic report of a patient's genetic test results, for both the family and the ordering physician. This model allows the ordering physician to have the opportunity to discuss test results with the family first, if they would like to do so. The Lineagen genetic counseling team is then available for post-test genetic counseling sessions to discuss the results and answer any questions that arise from the family and physicians.

To learn more about this model, I was able to listen to and discuss past telephone genetic counseling sessions with Lineagen's genetic counselors, and it has changed my perspective on the efficacy of telephone genetic counseling. First, telephone counseling allows the family to talk to a counselor from a place of comfort and control. Families seem to feel more comfortable asking questions and are more open when they are not sitting in a clinic room with a genetic counselor directly in front of them. In addition, many children with ASD or DD can be extremely challenging to prepare for outings and clinic visits. These families particularly appreciated telephone genetic counseling for the convenience of not having to alter their child's schedule and take them out of their familiar environments.

Finally, one of the most valuable things I have learned during this experience is the importance of knowing the differences among various CMA platforms. I had not realized how many CMA platforms are available, and how the results of each can provide different clinical diagnoses and impact the medical management of a patient. I now understand how certain CMA platforms can detect long continuous stretches of homozygosity, which may suggest uniparental disomy or regions that are identical by descent. These regions increase the suspicion for an imprinting condition or recessive genetic condition mapping to these regions, both of which can impact the future diagnostic testing strategy for a patient.

Overall, this experience has dramatically enhanced my genetic counseling training. I feel more confident in my knowledge about CMA technology, in my ability to interpret results, and in my critical thinking skills to work through complex psychosocial issues. From my unique experience at Lineagen, I know I will be more competent and capable as a future genetic counselor.

Licensure / Billing & Reimbursement

Coding Corner

What's the Right CPT Code for You?

By Pia Summerour, MS, CGC and Kaylene Ready, MS, CGC

The Coding Corner is supported by the Coding Subcommittee of the National Society of Genetic Counselors' (NSGC) Access and Service Delivery Committee and aims to assist NSGC members with the application and understanding of governmental regulations and guidelines regarding terminology and Current Procedural Terminology (CPT)/International Classification of Diseases (ICD) coding in genetic services as well as keep the membership educated regarding billing and reimbursement issues.

What's the best way to bill for your office visits? Unfortunately, there's no straight, easy answer. Since most genetic counselors are not taught about billing and coding in graduate school, this can be murky territory for us to navigate. But fear not – you have several options, such as billing for professional services (either via direct billing or "incident to" billing) and facility fee billing.

Professional Services

Direct Billing

Direct billing refers to a genetic counselor billing in his/her own name and National Provider Identifier (NPI) number as an independent provider. This option is not applicable to Medicare, as genetic counselors are not recognized by the Center for Medicare and Medicaid Services (CMS). State Medicaid programs could choose to cover more than Medicare, so it also may not necessarily apply to Medicaid. This type of billing is allowed in every state with licensure, but is dependent on local payer policies.

"Incident to" Billing

"Incident to" billing refers to a genetic counselor billing for his/her time in the name of a supervising physician. For Medicare this is the most appropriate way to be reimbursed, but there are several criteria that must be met, including that the genetic counselor and physician must be employed by the same entity and the physician must be present in the office suite or building during the consultation.

Facility Fee Billing

Another way to bill for your time is via facility fee billing. In order to bill a facility fee, you must be employed by the "facility" submitting the facility fee bill.

Common CPT Codes

Billing for professional services may require credentialing by your employer and/or local payers and utilizes the CPT coding system. The following are common CPT codes/categories:

- Evaluation and Management (E&M) Codes – require that certain key components be addressed or that more than 50% of time is spent counseling (4 categories):
  1) Consultation (99241-5) – requires a referral from a recognized health care professional
  2) Office visit (99201-5, 99211-5) – for self-referred patients
  3) Preventive medicine (99404-4) – for individuals without a specific illness or diagnosis for which the counseling might otherwise be used as part of treatment (these codes are distinct from E&M services that may be reported separately when performed).
  4) Health behavior and assessment (96150-5) – for individuals who have acute or chronic illnesses
- 96040 - created specifically for non-physician genetic counselors

If you are interested in exploring all of your options, we recommend that you meet with your facility's billing and compliance team.

For more information about billing and coding, we recommend the "Credentialing, Coding, and Compliance" course, which will be available for purchase by October 2012 on the NSGC website.

Additionally, the Payer Subcommittee recently administered a Billing and Reimbursement Survey to assess the needs and knowledge of the membership. Stay tuned for more targeted education based on the results of the survey!

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 ***Pia Summerour*** *(pia.banerji@utsouthwestern.edu) or* ***Kaylene Ready*** *(kaylene@counsyl.com)*

SIG Speak

From the Prenatal SIG

Non-Invasive Prenatal Testing: One Counselor's Preliminary Experience

By Katharine Coles, MS, CGC

I feel it is a safe assumption to make that most prenatal genetic counselors welcome the day when prenatal diagnosis can be offered without the risks currently associated with invasive chorionic villus sampling (CVS) and amniocentesis. When the first inklings about the clinical launch of non-invasive prenatal diagnosis/testing (NIPT) via cell-free fetal DNA were released, the prenatal genetic counseling community was anxious and intrigued. After several bumps in the road, this testing became clinically available in the U.S. in November 2011. Originally launched for the detection of Down syndrome, the conditions validated for NIPT to detect have recently expanded.

In some ways, NIPT should have been a dream come true. However, it was met with hesitation and resistance in the analytical genetic counseling world. Prenatal genetic counselors wondered, "How has this test been validated? To whom should we offer this test? How does it fit among traditional screening and diagnostic options? Will patients receive adequate pre- and post-test genetic counseling?" After several months of experience offering NIPT at our prenatal clinics in Houston, Texas, I feel it is time to reflect...

Guidelines from the initial laboratory offering NIPT stated that NIPT should be offered to a woman who is at "high risk." Therefore, we began incorporating this option into standard advanced maternal age (AMA), abnormal serum screen, abnormal ultrasound, and "significant family history" sessions along with maternal serum screening, CVS, and amniocentesis. My colleagues and I find genetic counseling for NIPT takes an average of five minutes when incorporated into a genetic counseling session. In some cases this can be as brief as thirty seconds, or as detailed as thirty minutes.

At this point, I would like to step away from my non-directive counseling role and provide a commentary on what actually went though my mind during my first few sessions offering this test:

"This feels confusing."

Offering NIPT seemed like wedging another option awkwardly between screening and invasive testing. For my general patients who were AMA and not overly anxious, it put too many decisions on the table. My patients with abnormal serum screens didn't want "just another blood test." My statistically minded, high-anxiety patients wanted *The*.

Most. Accurate. Option. What was a genetic counselor to do? This feeling validated original hesitations I had regarding NIPT. Some of the drawbacks that were extremely apparent in the first few sessions included:

1) **Newness.** Although these tests have been validated, they do not hold the historical power of first and second trimester screening, CVS, and amniocentesis. I cannot inform my patients based on *thousands* of validated cases; the current published numbers of affected cases identified by NIPT are only in the hundreds.

2) **Choices.** Many patients struggle to understand how this test is similar to, and different from their other options. Stating that something is "close to" the accuracy of an amniocentesis is a difficult concept, especially if a follow-up amniocentesis is recommended.

3) **Scope.** This test is simply not as comprehensive as the alternatives. A patient receiving a first trimester screen may only get a "risk value," but she also gets information from the nuchal translucency ultrasound that may imply a birth defect or information from her analytes warning of other potential adverse pregnancy outcomes. In our center, a patient choosing invasive testing will be offered a complete karyotype as well as chromosomal microarray (CMA), which could potentially detect far more than a limited number of chromosomal abnormalities.

4) **Payment.** Yes, always a concern. Our patients with Medicaid (or no health insurance) initially faced the whopping cost of almost $2,000 when NIPT was first available. This made offering the test feel insensitive. I had to be careful as to how/when this was presented to my patients without private insurance.

In all honesty, NIPT, you made things a little more confusing at the beginning.

And then…

A small group of women emerged from the fog. These women simply **would not consider** invasive testing. The risks for miscarriage were unacceptable to them. However, they were nervous about their screening results, ultrasound, age, and/or previous pregnancy histories. Although it is recommended that the results of NIPT be confirmed with a follow-up CVS or amniocentesis, this is not required. The majority of my patients who choose NIPT are looking for information and reassurance. In my experience, most would not consider termination.

Therefore, for this subset of women who are using further testing for information (and not for decision-making about continuing the pregnancy), NIPT has been a great option. Alternatively, couples who would consider CVS or amniocentesis frequently seem to be comfortable enough with the invasive procedures to skip NIPT altogether.

In addition to now finding a niche of patients that benefit from NIPT, some of my prior hesitations have been resolved. Recently, trisomy 18, trisomy 13, and in some platforms, monosomy X, have been added to NIPT. Some laboratories are offering coverage for Medicaid patients, and payment plans have been set up for uninsured/underinsured patients. I have had amazing success applying for financial assistance, and I encourage you to try! Additionally, NIPT has been heavily advertised and publicized, so some of the "newness" is waning. Patients are more regularly bringing this option to the table before I have a chance to introduce it. Some growing pains have worn away.

Yet, major limitations remain. At this time, we are still working through the best way to handle patients who want NIPT *before* they have a first trimester screen. As mentioned previously, the nuchal translucency ultrasound and maternal serum analytes have utility beyond detecting chromosome abnormalities. Is it worth our health care dollars to test these patients twice? Also, consider the confusion patients may face in the case of discordant screening and NIPT results. I also have hesitations for women who choose NIPT based on AMA or abnormal ultrasound findings. What may we be missing? I have completely come to terms with NIPT for Down syndrome if one screens positive for Down syndrome. This would accurately answer the question that has been raised. However, if one has a soft sign or ultrasound finding, this could indicate a condition other than the chromosome abnormalities tested for via NIPT. We are therefore potentially missing a chance to diagnose via complete karyotype or CMA if we do not offer invasive testing. Also, other aneuploidies are absent from the conditions tested by NIPT, so my patients who are AMA are missing out, too.

Obviously, these are limitations that are explored in a genetic counseling session, and I encourage you to explore them for yourself. I recognize that my personal experiences are based only on a limited sample of patients. I enlisted feedback from other prenatal genetic counselors in my area while writing this article and found that their experiences were similar to mine. However, our Houston population is likely different from that of other areas of the country and world.

For the many genetic counselors who have yet to delve into the world of NIPT, I hope to have offered some real-world perspective to illustrate how my initial response was both right on target, and an incredible misstep. Just as CMA, first trimester screening, quad screening (and even triple screening!) raised their own unique concerns when they were first introduced; these obstacles are the unavoidable truths of developing technologies. One day, we will likely see NIPT also become the standard of care.

NSGC News

2012 Jane Engelberg Memorial Fellowship Student Research Award Recipients

Editors' Note: *The Jane Engelberg Memorial Fellowship Student Research Award, now in its second year, funds up to $500 to promising research proposals developed by up to ten genetic counseling students per year. The proposals may be related to any area of research. The 2013 student proposal submission deadline is* ***June 1, 2013****. More information and application guidelines can be found on the National Society of Genetic Counselors' website at: http://www.nsgc.org/About/JaneEngelbergMemorialFellowship/tabid/464/Default.aspx.*

The following are proposal abstracts from the 2012 Jane Engelberg Memorial Fellowship Student Research award recipients, listed in alphabetical order by last name.

Heather Andrighetti (University of Toronto, Toronto, Ontario)

Parental causal attributions of obsessive-compulsive disorder and implications for genetic counseling: An exploratory study

Obsessive-compulsive disorder (OCD) is a relatively common anxiety disorder that poses a significant burden to affected individuals and family members. OCD has a complex etiology, with estimates of heritability for early-onset OCD in the range of 45-65%. Patient understanding of the causes of psychiatric disorders is instrumental in adaptation and coping in mental illness, and is known to influence help-seeking behaviors and orientation to treatment. Furthermore, genetic counseling for psychiatric disorders empowers patients and reduces self-blame, anxiety, and perceived stigma. While studies of etiological understanding and genetic counseling interest have been conducted for other complex psychiatric illnesses, including schizophrenia and bipolar disorder, no similar studies have been conducted among families affected by OCD. There are substantive differences between the psychiatric disorders that have been studied and OCD; specifically, the latter is an anxiety disorder, rather than a psychotic or mood

disorder, is more likely to have pediatric onset than other psychiatric illnesses, and has been shown to have significant and distinct impacts on family functioning. This study aims to explore perspectives regarding causal attributions of OCD and the potential value of genetic counseling among parents of children with OCD, in order to inform clinical practice for this population. A sample of fifteen parents of children diagnosed with OCD will be recruited through the British Columbia Children's Hospital OCD Clinic and Translational Research Program in Vancouver. Qualitative semi-structured telephone interviews will be conducted and audio-taped, followed by thematic analysis of transcripts. Interviews will follow guides adapted from previous studies on perceptions of genetic counseling among adults with bipolar disorder and their siblings. This study will be the first to explore perceptions of the growing discipline of psychiatric genetic counseling among families affected by OCD. Gaining insight into the educational and counseling needs of this population will mark the first step towards provision of effective and comprehensive genetic counseling services for OCD. Furthermore, these results may help inform future studies and practice guidelines for other anxiety-related or pediatric-onset psychiatric disorders.

Dayna-Lynn Dymianiw (University of British Columbia, Vancouver, British Columbia)

A longitudinal study of maternal depression symptoms after identification of increased risk for fetal aneuploidy

During pregnancy, women are routinely offered prenatal screening for fetal aneuploidy. A small handful of studies indicate that shortly after receiving "positive" test results that indicate their pregnancy is at an increased risk for aneuploidy, women often experience symptoms of depression/anxiety. However, it remains unclear as to whether or not these symptoms persist over time, and/or predispose women to postpartum depression. Therefore, it is our goal to conduct a longitudinal study to compare the trajectories of depression symptoms through pregnancy and the postpartum period between two groups of 100 pregnant women: those who receive prenatal screening results that indicate an increased risk for fetal aneuploidy, and those who receive "normal" prenatal screening results. Participants will be recruited by genetic counselors at the Provincial Medical Genetics Program, the British Columbia Prenatal Screening Program, and various physician and midwifery clinics in British Columbia. The women who agree to

participate will receive an online link to the consent form, demographic questionnaire and the Edinburgh Postnatal Depression Scale (EPDS). The EPDS will be administered at four-week intervals, from enrollment to three months postpartum. The information that we generate could be useful in providing a rationale for healthcare professionals to monitor the mental health of mothers who choose to have prenatal screening.

Theodora Jacobson (Case Western Reserve University, Cleveland, Ohio)

An assessment of the health, social, and daily living skills of adults with Down syndrome

Genetic counselors strive to provide a "balanced" description of Down syndrome (DS) to patients, so that patients have information upon which they can make autonomous decisions either prenatally or postnatally regarding their pregnancy or child. The National Society of Genetic Counselors has published guidelines for genetic counselors and other healthcare providers regarding key information to convey to families about DS, including information about how DS occurs and is diagnosed, medical complications, intellectual abilities, available resources, and information that demonstrates what a potential life is like for an individual with DS.[1] However, the guidelines' authors admit that there is a lack of data regarding the long-term natural history of adults with DS.[1] As adults with DS continue to live longer due to improved medical care, more current, up-to-date information would be useful so that genetic counselors can better describe the potential capabilities of this population. The purpose of this study is to assess the health, social, and daily living skills of adults with DS in order to provide genetic counselors with more accurate information to offer to families. Parents and primary caregivers of adults with DS over the age of twenty years from four organizations will be asked to participate in an online questionnaire adapted from a survey of individuals with DS in Italy.[2] It is hoped that findings from this study will provide updated information about health, social and life skills of adults with DS and help portray a more realistic, in-depth picture of those life skills, thus contributing to a "balanced" description of DS. In addition, this study strives to assist those who interact, either personally or professionally, with adults who have DS to gain a better understanding of this group's abilities.

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Sahil Kejriwal (Stanford University, Stanford, California)

A medical and psychosocial needs-assessment of families that have individuals affected by Duchenne muscular dystrophy in Madurai, India

The Indian healthcare system is still far from efficient at dealing with the complex needs of individuals who have Duchenne muscular dystrophy and their families. Additionally, the government and much of society are unaware of these needs. The aforementioned factors, combined with poor access to affordable health insurance schemes, results in numerous unmet medical, psychological and social needs that require attention. In order to elucidate these needs, interviews will be conducted with parents of twenty individuals who are affected by Duchenne muscular dystrophy in Madurai, India. The families will be chosen based on variables such as age, number of affected individuals, socioeconomic status, and literacy levels in order to reveal various circumstantial needs. The study will be carried out in collaboration with an advocacy/support group known as the Muscular Dystrophy Foundation of India, Madurai; the participant pool will consist of parents registered with this organization. The findings will be disseminated to stakeholders that can help meet these families' needs, such as community service clubs, clinicians, policy makers, and other advocacy/support groups.

Lesli Kiedrowski (University of Michigan, Ann Arbor, Michigan)

Exploring parents' understanding and utilization of variants of uncertain clinical significance identified from chromosomal microarray: A qualitative study

Chromosomal microarray analysis (CMA) is now a first-tier diagnostic test for individuals with unexplained congenital anomalies or developmental disabilities. Copy number variants (CNVs) identified through CMA may be implicated as the cause of health conditions, which can provide benefits both psychosocially and for medical management. However, many CNVs are benign polymorphisms with no confirmed health implications. Due to the rarity of pathogenic CNVs and rapid advances in technology and clinical implementation, there are large gaps in knowledge regarding abnormalities' phenotypic effects; it is not uncommon for CMA results to be classified as variants of uncertain clinical significance (VUS). This presents a family with ambiguity as to whether or not the result explains their child's condition, leaving them with a functionally uncertain diagnosis. Parents' expectations of, and desires for, a diagnosis and information to guide medical management may lead to them viewing uncertain findings as pathogenic; the unclear test results could also be dismissed as unrelated. These personal interpretations may affect not only parents' abilities to cope, but also their motivations to follow up with the genetics clinic and other referrals. Semi-structured qualitative interviews with parents of children who have received a VUS CMA result will be conducted to explore ways in which parents interpret and utilize their children's VUS genetic test results.

Whiwon Lee (University of Minnesota, Minneapolis, Minnesota)

Role of anxiety in genetic counselors' risk for compassion fatigue

Compassion fatigue (CMF) is a state of detachment and isolation experienced by healthcare providers who interact with patients in distress. Research suggests that 26% of genetic counselors are at high risk of experiencing compassion fatigue and another 57% are at moderate risk.[1] Also, approximately one in four genetic counselors has considered leaving the field due to CMF.[2] These studies have begun to identify personality traits associated with CMF risk, but, to date, none examines whether anxiety predisposes genetic counselors to CMF. A recent investigation of the relationship between trait anxiety (defined as "relatively stable differences in anxiety-proneness") and state anxiety (defined as "subjective feelings of tension, apprehension, and worry") and genetic counseling students' graduate program experiences found that genetic counseling students, on average, had high trait anxiety, significantly higher than typical adult working women and medical students.[3,4] This is concerning since anxiety can interfere with memory, decision-making skills, and overall performance, thereby negatively affecting one's functioning, sense of adequacy as a professional, and career satisfaction. Given the potential deleterious effects of anxiety on professional functioning, the purpose of the proposed study is to assess state- and trait-anxiety levels of practicing genetic counselors and to determine the relationship between anxiety and CMF risk. The findings will help to identify those individuals at highest risk of CMF, potentially explain why some genetic counselors could be at risk of leaving the profession, and help to inform continuing education programs for managing anxiety and CMF reactions.

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ABGC Update

The New Genetic Counseling Accreditation Body: An Overview of the Accreditation Council of Genetic Counseling (ACGC)

By the ABGC Board of Directors

Several years ago, the American Board of Genetic Counseling (ABGC) Board of Directors were advised by legal counsel that it is a conflict of interest for the certifying body to also accredit training programs. With this in mind, the ABGC has been moving towards a formal separation between the credentialing body (which will retain the name the American Board of Genetic Counseling) and the accrediting body (which will be called the Accreditation Council of Genetic Counseling or ACGC).

We are continuing with the final stages of planning and preparing for the official launch of the separate accrediting body for genetic counseling education and training programs. This article will review the structure and current status of the new ACGC organization.

Purpose of the ACGC

The ACGC shall provide leadership by establishing standards for graduate level genetic counseling education in order to protect the interests of students, the public and the integrity of the genetic counseling profession. More specifically, the organization will:

- Evaluate education programs to ensure compliance with the standards, and
- Accredit genetic counseling training programs that meet the standards established by the ACGC

Board of Directors

We are transitioning to the official composition of the ACGC Board of Directors (BOD), which will be comprised of nine board members, six of whom are certified genetic counselors, two educators or faculty members of genetic counseling training programs, and one public member. Board members will serve a three-year term and are eligible for one consecutive term.

The charter members of the ACGC BOD are the five elected ABGC Board members who have served on the ABGC Accreditation Committee, and four additional members who

were appointed to serve on the Transition Task Force and then agreed to continue on as charter Board members.

The charter ACGC BOD members are:

Holly Peay, President
Janice Berliner, Past President
Lisa Amacker North, Secretary/Treasurer
Sarah Noblin, Accreditation Review Chair
Robin Bennett, Transition Director
Cecelia Bellcross, Director
Jennifer Fitzpatrick, Director
Kathy Valverde, Director
Meredith Weaver, Director

Committees

There are seven standing committees for the ACGC that include Nominating, Program Review, Site Visitor, Standards, Finance, Grievance, and Executive.

Current Status of the ABGC and ACGC

It is anticipated that ABGC and ACGC will be operating as two separate organizations by January 1, 2013. The ACGC board and its Transition Task Force have spent the past year reviewing processes and procedures, developing bylaws, updating the Accreditation Standards, and collaborating with the ABGC to update its Practice-Based Competencies. The Articles of Incorporation for the ACGC have been filed with the appropriate offices of the Secretary of State and have been accepted. Additionally, the ACGC has been issued an employer tax identification number. These steps are required to form any new corporation. An application for nonprofit tax status is being prepared, and will be filed in the near future.

The ACGC is in the process of finalizing a logo and collateral materials. Additionally, we are working on a contract for management services. The Board felt that stability during this transition to two separate organizations was critical to success; therefore, at least for the short term, management services will continue to be provided by Applied Measurement Professionals, the ABGC's current management firm.

Please attend the ABGC Business Meeting at the NSGC's Annual Education Conference in Boston in October to hear the latest news and updates!

Global Genetics

To the Emerald Isle and Back: The Story of a Temporary Dubliner

By Karen Siklosi, MGC, CGC

In July of 2008, my fellow genetic counseling classmates were beginning their first jobs in the field; I was packing my bags and selling my furniture. My passport was current, I had Euro in hand, and there would be no familiar faces when my plane landed in Dublin.

Instead of entering the workforce (and finally getting a salary after nineteen years of full-time schooling!), I was on my way to Ireland for a year of research related to cystic fibrosis. My arrival there was the perfect culmination of equal parts effort and luck, beginning with the seemingly unrealistic idea of "I'd like to live in Ireland," which struck me sometime during the summer before my second year of graduate school. Having not studied abroad in college, I was interested in spending time in another culture, but was limited to English-speaking countries, thanks to eight years of studying Latin.

A few months later, I reached out to turn this seemingly unrealistic idea into something. At the time that I began looking for opportunities, there were five genetic counselors for all of Ireland and it quickly became clear to me that a new hire from the U.S. was not in the cards. So, I narrowed my focus to the disease that had first sparked my interest in genetics: cystic fibrosis (CF). I had previously done molecular work related to the *CFTR* gene in college, but turned to genetic counseling because I was interested in patients themselves – not just their DNA.

I contacted the Cystic Fibrosis Association of Ireland and offered to do anything – research, intern, fundraising, etc. Luckily, I received a response and was put in contact with Dr. Ed McKone, a respiratory consultant at the adult referral center for CF, located at St. Vincent's University Hospital in Dublin. He would be happy to host me as a researcher, provided that I came with my own funding.

After weeks of developing a project and proposal, I applied for funding through a U.S. Department of State Fulbright grant, which is a competitive program that sponsors research, education, or teaching in nearly any country. Fittingly, I received notification

that I had received one of three Irish grants on St. Patrick's Day in 2008. It was official: in late summer, I was moving to Dublin.

In Ireland, CF occurs more frequently than in any other country, with an approximate incidence of 1 in 1,461 births and a carrier frequency of 1 in 19. At the time I was there, the estimated age of survival was well below the U.S. average. Although it would be impossible to pinpoint one cause for the lower survival rate, a report issued in 2005 by an independent review of CF services in Ireland indicated that a lack of appropriate inpatient facilities was likely a contributing factor. Patients, families, and others within the CF community had become increasingly vocal about the lack of appropriate facilities – particularly, they claimed, because the government and the Health Service Executive (the country's provider of national healthcare services to every Irish resident) had been promising better CF facilities for several years and those promises had gone unfulfilled.

Because of the abundance of stories about CF in the news, my proposal to research the condition was received with open arms. Although I had originally offered to work on any of the projects already underway, Dr. McKone suggested that I design my own under his guidance, since he had never had someone with a genetic counseling background work with him. We created a survey-based project that would assess the knowledge, attitudes, and education of adult patients with CF and their parents. Additionally, I collected clinical information about each patient. I hoped to correlate level of CF knowledge with clinical outcomes, as well as identify areas where education about the condition may be improved.

I approached over 150 patients and parents to help me create, validate, or complete my survey. Nearly everyone was willing to help or participate, and I was able to complete my project within my ten months of funding; results were published the following year. Working at St. Vincent's was a joy. The CF team and patients were more than welcoming, which is something I wasn't expecting. They asked me about genetic counseling, why I was interested in CF, and – perhaps most frequently – how I was enjoying their country. The inpatients, many of whom spent multiple weeks in hospital with little entertainment, were particularly eager to chat with me when I stopped in. I learned that it can be hard to avoid getting attached to patients with a chronic disease; seeing them every day, talking about their lives, and hearing their stories can seem like visiting with a friend.

Outside of work, I drank in the Irish culture. I took dance classes for the first time in years (taught by one of the original *Riverdance* performers, no less!) and ran my first half-marathon. I (very unsuccessfully) attempted to learn the Irish language, still spoken in areas called the Gaeltacht, and fell in love with pub culture, which involves a pint, good friends and conversation, and perhaps even live music.

I was fortunate to present some of my work at both the National Cystic Fibrosis Conference and the Cystic Fibrosis Association of Ireland meeting and also had the opportunity to attend several government-related events because of my affiliation with the Fulbright group. Upon my return to the U.S., where I began work as a pediatric

genetic counselor, I joined a group within the CF team at my institution working to create a formal transition process from pediatric to adult CF care. I now work in a research role on several different projects related to CF at Johns Hopkins University and have surprised myself with how often I relate something to my experience abroad.

I am particularly grateful for the advice and guidance of two key people: my Program Director, **Shannan DeLany Dixon**, **MS**, **CGC**, for not letting me give up on my idea of living in Ireland, and to Dr. Ed McKone for his willingness to take me on as a mentee. Although moving alone to a new country had its ups and downs, I cannot think of another experience that was more helpful or humbling.

For more information about the Fulbright program, please visit http://www.iie.org/fulbright/.

*If you are or know of a genetic counselor with an interesting international story, please contact **Janice Berliner**, column editor, at berlinej@mskcc.org to discuss submission of an article.*

Student Forum

"So I have some bad news…"

By Sarah Witherington, Center for Genetic Medicine Program in Genetic Counseling at Northwestern University, Class of 2013

***Editors' Note*:** We are pleased to welcome **Sarah Witherington** to the *Perspectives in Genetic Counseling* team as our new Editor for "Student Forum," and we invited her to introduce herself to all of us by writing her first column.

For many genetic counselors, a good bit of our time is spent reflecting on giving bad news to patients. In fact, we have entire books on how to break bad news in the "best" way possible. I've thought about how frequently we have to say to our patients, "We got the results back from your child's microarray and I have some difficult news," or "We received your *BRCA* tests results and the results weren't what we were hoping for," or some variation of those words. How many times have I heard or said those words during a session in clinic or during a phone call disclosing test results?

For most of us, it isn't very often that we are on the receiving end of those statements. Honestly, I had never spent much time thinking about how I would react in such a situation. So when my dermatologist's nurse called recently and started with, "Are you available to talk?" then, "I have some bad news…" and ended with the word "melanoma," I was sent into shock. At age 24, I had been diagnosed with melanoma.

After the word "melanoma," things became a blur. I remember hearing something about a surgery and needing to schedule an appointment right away, but I don't remember much else. I later looked at how long the conversation between the nurse and I had lasted, curious to see how much of the information I had lost. The conversation had lasted for around five minutes and somehow all I had heard were the words "melanoma," "surgery," and "appointment" (which thankfully I had managed to schedule through my shock). Reflecting back, it's no wonder that it's rare for our patients to remember much of what we tell them.

Although I may not recall much of what the nurse said, I will likely never forget how I felt. My heart stopped when she asked if I was available. My stomach sank when she told

me I had melanoma. My heart sped up and panic set in right around the time she mentioned surgery. As genetic counselors, we are trained to be good listeners, to reflect on our emotions, and to be aware of our coping mechanisms. I am supposed to have specialized training and all I can remember from my bad news conversation is a few words, a sinking feeling in the pit of my stomach, and some moments of panic? I can now imagine the disadvantage our patients must have when receiving bad news.

As I sat quietly in the waiting room before surgery, I tried not to think about how much of my skin and underlying tissue they were going to have to take to fully remove the cancer. I tried to use one of my coping mechanisms, humor, to lighten the tension in those around me, even though I was still terrified inside. As I went into surgery, I joked with my surgeon that I could no longer win the title of "Miss America" since my shin would soon look like something out of "Bride of Frankenstein." She looked at me and laughed, saying, "No, but you can be 'Miss Melanoma.'" I immediately felt better knowing that my surgeon could joke with me even while going into a very serious situation. Humor may not work for everyone, but it helps ease my anxiety so that I can face a daunting situation head on.

There is now a three-inch incision and a nice divot in my shin from where they removed the underlying tissue that is still slowly healing. I am happy to say, however, that I am currently cancer free, even if it means that I can no longer win beauty pageants that I was never actually going to enter.

Before that initial phone call, I sometimes found myself feeling annoyed when patients would come back to clinic after a diagnosis and claim to remember nothing from the session. At the time, I couldn't imagine spending 15, 20, or even 30 minutes talking to someone and not being able to recall anything said, especially in such a serious situation. Now I have more compassion for these lapses in memory since I know I am just as "forgetful" as other patients. It's important to remember the impact we have on a patient's life every time we have contact with them, whether it is in person, over the phone, or though written communication. While I wish this were something I didn't have to experience, I am thankful for the change in perspective that this diagnosis has given me. It is my hope that I am more empathetic with my patients, and can use what I have learned from this ordeal to be a better genetic counselor.

I have been taught and have learned so many things over the past year, and I will always be grateful for the training I received at Northwestern. I am also appreciative of the opportunity to join the team at *Perspectives in Genetic Counseling*, not only to share my story, but to help other students share theirs as well. It is my hope that this article will allow you to reflect on how you give and react to bad news both professionally and personally, and of course, to check your skin periodically and to visit a dermatologist.

The New Graduate Life

Shifting Gears and Finding Direction

By Chris Colón, MS

***Editors' Note*:** We are pleased to announce **Christine (Chris) Colón** as our new Editor for "The New Graduate Life." Chris is transitioning to this role from a previous one with the *Perspectives in Genetic Counseling* team, and we thought it fitting for her to write her first column.

As any new graduate can tell you, after completing your education, life is different. There is often a mixture of overwhelming emotions: the relief of being finished with school, sadness from no longer seeing classmates as often, excitement at the prospect of starting a career, and the accompanying fear that you will not find a job. There may be a seemingly endless waiting period until the next thing comes along: viable employment, the beginning of further academic study, or a big event in personal life, like relocating, getting married, or starting a family. Life after school is a period of transition, and one that can be wonderful, unsettling, and confusing – all at the same time.

In May 2011, I had just completed my training program, and was happy to be finished with school. I was also totally unsure as to what would come next. I knew that I wanted to find a job, so I set about the task by updating my résumé and building a list of references. I applied to what few jobs I would find advertised, and often ran into classmates on interviews. I became anxious when weeks passed without new job postings or a follow-up call from employers. I became consumed with my lack of employment, and increasingly concerned about losing skills I had worked so hard to gain. After several weeks of getting nowhere, I decided to change direction. If I was unable to find a paid position, I realized I could retain my skills (and possibly learn new ones) if I spent some time volunteering in a field that utilized genetic counselors and focused on patient care.

To be honest, I wasn't crazy about the idea of working without receiving a paycheck; there is a lot to be said about being compensated for your time and effort. Still, in my mind, unpaid experience seemed better than no experience, and being busy would keep my mind off being unemployed. After making the decision to volunteer, my first step was to figure out where I wanted to donate some of my time. I wanted my experience to be

both valuable and stimulating, so I began thinking about my personal and professional interests (and where my energy could be usefully expended). The first thing that came to mind was a unique internship placement I had in September 2010.

Part of the genetic counseling training program I completed includes an annual lecture on teratogenic agents, outlining how to counsel on the risks for various exposures during pregnancy. It was at that time that I first met **Sharon Lavigne**, **MS**, a genetic counselor and Coordinator of the Connecticut Pregnancy Exposure Information Service based out of the University of Connecticut. During the lecture, our class was introduced to the Organization of Teratology Information Specialists (OTIS), a group of professionals with expertise in teratogen counseling that could serve as a resource for our future patients. In 2010, I completed an internship at the University of Connecticut, where I had the chance to work with Ms. Lavigne on preconception and prenatal cases of teratogen exposure through in-person and phone counseling. By the time I was finished, I had developed a strong interest in how exposures affected women before, during, and after pregnancy. It was this interest that prompted me to contact Ms. Lavigne again, and we discussed ways in which I could become involved with this specialty.

I joined OTIS and was placed on the Education Committee, a subgroup of the organization that works to ensure that OTIS continues to help educate patients, providers, and the public about teratology and birth defects. They have several ongoing responsibilities, such as creating new fact sheets, evaluating current fact sheets for content and language, and conducting recent literature reviews on topics in need of updates. The Committee members, who at that time I had met only through email, were encouraging and supportive as I tried to find ways to contribute. Eventually, I became acquainted with the correct style and format of the material, and was able to provide some useful suggestions. It felt great to have my feedback appreciated, and I gained a better understanding of how to counsel patients on their potential exposure-related risks.

After a few months, the Education Committee began planning their presentation session for the OTIS Annual Meeting. When it was decided that the focus of the presentation would be the importance of health literacy, I was very excited. My Master's capstone project was focused in part on evaluating genetic health brochures to ensure as many patients as possible could read and understand the information. Having firsthand knowledge about the topic allowed me to feel comfortable enough to share my ideas and participate in the creation of the presentation. It felt good that I had something to offer to the group. I worked closely with another OTIS member, Joselyn Hall, MPH, to create a presentation for the Annual Meeting. Our aim was to create something that was factual, relevant, and not too boring. After our work was reviewed and approved by the other Committee members, I was asked to give the presentation to the attendees. I was so nervous about the idea of speaking in front of the organization I had only recently joined that it took me a week to agree to do it. Finally, I decided to just go for it – if nothing else, I could count it as a learning experience.

I attended OTIS's 25th International Conference (their Annual Meeting) in Baltimore, Maryland this past June. In addition to the interesting and informative sessions and

workshops, it was also a great opportunity to meet some of the experts I had communicated with through email and finally match up faces and names. The group was so friendly and welcoming that my nervousness began to subside and I started to feel at ease. On the final day of the meeting, I delivered the presentation on health literacy with confidence, and I'm pleased to say it was well received by the attendees.

During the drive home from the meeting, I had some time to reflect on the year since graduation. I was surprised at how far I have progressed in that short period of time. I am no longer a new graduate that is unsure about how I fit in to the field of genetic counseling. Participating in both paid and unpaid work opportunities has allowed me to become more confident in my own abilities and to more easily identify where I can improve. I am excited to find new occasions to work with other professionals and learn from their varied experiences and, although I cannot be sure where my career path will ultimately take me, right now I feel like I am moving in the right direction.

Genetic Counselor Publications

Feature Article

By Sara Spencer, MS, CGC

Basile JE, Donnenfeld AE. The importance of a genetic evaluation of prospective ovum donors: A study of family history and genetic carrier testing. *J Reprod Med*. 56(9-10):415-20. 2011.

Joyce Basile, MS, CGC

Most prenatal genetic counselors have found themselves in this pickle before. You begin a session as usual and then the patient informs you that she used either an ova or sperm donor, or both. When you enter into your discussion about family history and carrier screening, your standard discussion comes to a halt as you attempt to gather the limited family history information available about the gamete donor(s), and ponder whether or not to offer carrier screening to the biological parent, if available. Some questions circulating through your mind may include: Will the donor be available for carrier screening if the biological parent is found to be a carrier of an autosomal recessive condition? Had the donor been screened by family history or with genetic testing as part of the donor screening process? With roughly ten percent of *in vitro* fertilization (IVF) cycles in the United States (U.S.) involving the use of an ovum donor, this situation is not exceptional in our field[1].

Genetic counselor **Joyce Basile** and her mentor, perinatal geneticist Alan Donnenfeld, have thought about this interesting quandary in more depth, and have performed a retrospective research study exploring the carrier testing performed, the results of the carrier testing, and family histories of ova donors. In Basile and Donnenfeld's article, they review the current guidelines for gamete donors set by the American Society of Reproductive Medicine (ASRM). These include some specific guidelines regarding cystic fibrosis carrier screening and family history assessments, along with some general guidelines regarding other ethnicity-based carrier screening and chromosome analysis.

The current featured article supports the need for a genetic counseling evaluation of women who volunteer to donate ova. Basile explained, "Currently, there is little

consensus about the type of genetic evaluations and/or carrier screening these potential donors should undergo prior to donating ovum. At this time, most carrier screening of these donors for genetic disorders is performed at the discretion of the fertility center, with limited guidance from the American Society of Reproductive Medicine (ASRM) regarding recommendations for some genetic testing and family history evaluations. This retrospective project evaluated prospective ovum donors who had undergone genetic counseling to determine whether they had a family history that might preclude them from being a suitable donor. Upon reviewing the ovum donor's family history through pedigree analysis, we found that 210 (22.1%) of 950 potential oocyte donors had at least one fetal risk factor [discovered by family history assessment] such as mental retardation, cystic fibrosis, and hemophilia." Basile and Donnenfeld also studied the laboratory results for disorders such as cystic fibrosis, Tay-Sachs disease and fragile X syndrome for patients who had undergone genetic carrier screening for the referral indication of egg donor screening. They determined that 15 (6.1%) of 244 prospective donors were found to be carriers of hereditary diseases, which could pose an increased risk to a fetus.

Basile stated, "The use of a genetic counselor to aid in the screening process of potential ovum donors can help to quantify the genetic risks associated with a pregnancy that may be conceived from a particular donated egg. This may help to reassure women who attempt to conceive using an anonymous donor."

When asked how she thinks genetic counselors can be integrated into the gamete donor process, Basile responded, "In my opinion, the genetic counselor can be an invaluable member of the team who evaluates prospective gamete donors (male or female). A genetic counselor is highly qualified to provide pedigree risk assessment and discuss options with the referring physician for appropriate genetic testing [based upon ethnicity and family history risk factors]. The genetic counseling may be provided in person or sometimes via telephone. While there may be different opinions regarding the appropriate time in the evaluation process for the genetic counseling appointment, it seems to me to be most reasonable to provide this service once the patient (ovum or sperm donor) has passed the preliminary evaluation by the reproductive endocrinologist."

Basile graduated from the Beaver College (now Arcadia University) genetic counseling program in 1998, is a prenatal genetic counselor at Integrated Genetics, and has been in this role since 1999, beginning her career in the Philadelphia/New Jersey area and then transferring to San Antonio, Texas in 2007. Like many genetic counselors, Basile's job responsibilities consist almost entirely of providing clinical genetic counseling. Given that her job responsibilities are mostly clinical, she manages to find the time and motivation to formulate research questions, perform research, write manuscripts, publish, submit research abstracts, and present posters with the help of a mentor. She stated, "I also work with a fabulous physician, Dr. Alan Donnenfeld, who is a wonderful mentor and supporter of my interest in research and publication. He encouraged me to combine two of my poster abstracts in this journal manuscript, and he guided me through the writing and submission process."

For those genetic counselors or new graduates interested in contributing to research in our field, Basile has some step-by-step advice for you! She stated, “I think the two most important tips are to identify a question that you would like to answer, and then find a mentor who will help and motivate you. Next, you evaluate what you need to do to develop an answer to that question.”

References

1. Society for Assisted Reproductive Technology: National summary of IVF success rates, 2009. (https://www.sartcorsonline.com/rptCSR_PublicMultYear.aspx?ClinicPKID=0).

AEC Update

NSGC 31st Annual Education Conference

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

Something for Everyone

By now, you should have received your official conference booklet detailing the National Society of Genetic Counselors' (NSGC) 31st Annual Education Conference (AEC) and we hope that you have already made plans to attend. The conference will be held **October 24-27, 2012** in **Boston, Massachusetts**. The NSGC membership has grown significantly, both in the number of members and the variety of professional environments in which we practice. To celebrate our diversification and growth, we will be in a conference center venue for the first time this year. Take a sneak peak at our space by visiting:
http://advantageboston.com/hynes/floor-plans-and-specifications.aspx

Plan Ahead for Boston: Download the NSGC AEC App Now

Scan the above QR code or search "2012 NSGC AEC Mobile App" in the iTunes or Google Play Store to download your free AEC app. Scroll through the "Schedule at a Glance" feature and star the sessions you plan to attend. Selected sessions then appear in your "My Schedule" section. Browse the speakers and the featured sponsors. Plan your evenings and meals in Boston by visiting the "Local Places" section. You can also link through to the NSGC's Twitter feed and Facebook page from the App.

Further technological advances to expect in Boston include being able to use a QR scanner from your smartphone to access session information on site, and having free Wi-Fi access in the conference center to facilitate tablet, netbook, and laptop use. If you do not already have a QR app, consider downloading one prior to arriving in Boston. For

example, RedLaser has a free QR scanner available in iTunes for iPhone and iPad users; similar free apps are available for Android, BlackBerry, and other smartphone users. For those that would prefer to download or print conference handouts in advance, a link to the conference handouts will be sent to registered attendees prior to the conference.

Educational Offerings Abound

The 2012 AEC will begin on Wednesday with the "Welcome to the AEC" orientation at 2:00 p.m., followed by the opening Plenary sessions at 3:30 p.m. including the Janus Series, Best Abstract Awards, and Natalie Weissberger Paul and Audrey Heimler Special Project Award announcements. Concluding this kickoff will be the Welcome Reception in the Exhibitor Suite on Wednesday evening. There will be three full days of outstanding educational opportunities on Thursday, Friday, and Saturday with the conference concluding at 3:30 p.m. on Saturday.

Over the course of the conference, attendees can earn up to 3.23 Continuing Education Credits (CEUs) by attending educational sessions and sponsored events. Attendees wanting to maximize their learning and CEU opportunities should consider attending a Pre-Conference Symposium Wednesday morning for an additional 0.5 CEUs. The Pre-Conference Symposia offer high level, in-depth sessions for specific specialty practice areas, new issues in genetics and genomics, or professional development topics. Please note that Pre-Conference Symposia require separate registration from the AEC and will have limited space available. Sign up early!

Outreach in Boston

In an effort to reach out to the community of our host city, the NSGC annually conducts an Outreach Event during the AEC. **Katherine (Kat) Lafferty** and the Outreach Committee are coordinating this year's event. They are already hard at work creating a PowerPoint presentation entitled "Genetic Counseling as a Profession" to present to college students in New England and are developing a Facebook page (http://www.facebook.com/GCcareerevent). The students who attend these presentations have been invited to join us Friday afternoon during the AEC to attend educational sessions and to hear a panel discussion of genetic counselors from a variety of job experiences and work settings. If you are interested in being on the panel to discuss your career in genetic counseling, please contact Kat at Katherine.lafferty@gmail.com.

Accommodations

The Hynes Convention Center is located in the heart of Boston's historic Back Bay neighborhood. Two nearby hotels, the Boston Marriott Copley Place and the Sheraton Boston Hotel, have room blocks available for NSGC AEC attendees. Additional rooms

are added to the NSGC block as they become available. Please continue to check with the NSGC hotels for availability. Surrounding the convention center area, attendees will find world-class shopping, dining, and entertainment. Visit http://www.cityofboston.gov/visitors/thingstodo.asp or http://advantageboston.com/Hynes/Advantages.aspx for more information.

Many Thanks

We would like to thank our Conference Subcommittee members for all of their time and efforts in the planning of the 31st AEC: **Mary Jarvis Ahrens**, **Julie Culver**, **Katie Dunn**, **Patricia Devers**, **Lori Erby**, **Lauren Hache**, **Brandie Leach**, **Anne Madeo**, **Catherine Vendola**, **Meredith Weaver**, and **Emily Windsor**. An endeavor such as the AEC does not come to fruition without the dedication of the member volunteers and our Executive Office staff. We would also like to thank **Leigha Senter Jamieson** and **Kelly Jackson**, Education Committee Chair and Vice-Chair, **Katherine Lafferty**, Outreach Coordinator, and **Janet Williams**, NSGC Board of Directors liaison, for their guidance during the planning for the 31st AEC.

Safe travels and we look forward to seeing you in Boston!

Claire N. Singletary (Claire.n.singletary@uth.tmc.edu), 2012 NSGC AEC Chair
Quinn Stein (quinn.stein@sanfordhealth.org), 2012 NSGC Vice Chair

Resources / Book Review

Reviewed by Lara Reichman, BSc

Saving Henry: A Mother's Journey

Author: Laurie Strongin
Publisher: Hyperion (February 22, 2010)
Pages: 224
Retail price: $29.50
ISBN: 978-1401323561

From the first page of Laurie Strongin's Saving Henry: A Mother's Journey, it is clear there is immeasurable love driving her determination to evade the fatality of her son's Fanconi anemia (FA) diagnosis. Strongin details the balancing act she and her husband attempt in trying to lead a "normal" life, while making every effort to find a perfect human leukocyte antigen (HLA) match for a lifesaving bone marrow transplant for their first child, Henry.

Strongin chronicles her journey in discovering Henry's diagnosis soon after birth and the ensuing news that he will need a bone marrow transplant by the age of five. Armed with this knowledge, finding a suitable donor becomes the only focus for Strongin and her husband. Having planned to have more children, though not necessarily so close together, they decide that if there is hope that another child could be a bone marrow donor for Henry, they are ready to have that child as soon as possible. Before pre-implantation genetic diagnosis (PGD) was an option in their quest for a matched donor, Strongin and her husband have another child.

While free of FA, this child is not an HLA match. During this pregnancy, they are informed of possible clinical trials of PGD beginning for FA and for HLA matches and set off on an emotionally, physically, and financially draining mission. The first couple in the world to try PGD to find an HLA match for FA, Strongin and her husband find themselves in a unique ethical position, becoming the focus of public scrutiny for discarding embryos that are not HLA matches, but are otherwise predicted to be normal.

Repeated throughout the book is the sentiment Strongin articulates as, "I believe in love and science, nothing more and nothing less." Her love for Henry and her family is evident through her storytelling. She describes in painfully honest detail the struggle between placing hope on science, and coming to grips with its limitations when undergoing nine PGD cycles without a successful pregnancy. She brings to light the self-doubt and personal responsibility that is sadly associated with genetic diseases – the feeling that parents have unknowingly brought great pain to their child's life, and the helplessness that comes with a genetic diagnosis that cannot be undone. However, she

counters this with an attitude of seizing the moment and living each day to its fullest, an outlook particularly inspirational given the severity of FA.

Though Henry ultimately succumbs to an infection, the title of Strongin's story is still perfectly accurate. Saving Henry is about just that, the unwavering love a mother has for her child and her resolve to do everything possible to free him of his illness. This makes Saving Henry a book accessible to nearly anyone. For people who are trying to better understand what it means to parent a terminally ill child, Strongin's narrative illustrates the daily struggle of remembering the diagnosis and temporarily putting it aside, choosing not to live in its shadow. Her description of her *in vitro* fertilization (IVF) cycles also showcases the struggles rarely discussed with building a family, and could be beneficial to other women and families using assisted reproductive technologies.

Henry's story is a crucial read for any genetic counselor because it serves as a reminder of the roles we play for patients. Strongin shows mastery of difficult and confusing scientific terms and explains them throughout the book, making it accessible to readers of any level of scientific literacy. These definitions also highlight the extent to which parents and patients must learn new terms when given a diagnosis, and how genetic counselors can make that learning process less isolating. Moreover, Strongin's understanding of these concepts shows the constant determination on the part of parents to push themselves to grasp as much of the diagnosis as possible, in an effort to seek out every opportunity for treatment and management. This focus is to be commended, and genetic counselors are in a unique position to recognize it and complement it, hopefully making "a mother's journey" even the slightest bit less difficult.