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President's Beat

Rejuvenation and Inspiration at the Annual Education Conference

I can hardly believe summer is over, and many of us will soon convene in Dallas for our Annual Education Conference (AEC). Throughout my career, I've looked forward to the AEC for more than an opportunity to learn. Of course, many of the reasons I attend the AEC are those you might find on the brochure: updates on the old standards, awareness of emerging clinical trends, networking, and getting those continuing education units (CEUs)! But the AEC is much more to me than just a collection of CEU certificates or note-laden handouts. It's my inspiration and time for rejuvenation.

The AEC is our one opportunity as genetic counselors to come together in person, celebrate our success, and toast the future. I've found it hard to explain to my friends and family how the NSGC conference is different from other professional conferences, more than a sum of its parts, but actually a supply of collective energy that reinvigorates my passion for advancing the field. They don't understand because they aren't genetic counselors who hold the solutions to many of the challenges facing genomic medicine, and they don't understand why I seem to forget their phone numbers while I'm away for almost a week!

This year's AEC will not disappoint. Thanks to the AEC Subcommittee, an exciting program awaits. You can view the agenda at www.nsgc.org/conferences/aec.cfm. I'm especially excited about a few presentations in particular: the Professional Issues Panel on Saturday morning, the late-breaking session on Sunday afternoon, and the Incoming Presidential Address by President-Elect **Karin Dent**, who will also speak on Sunday afternoon.

Many NSGC members rate the issue of reimbursement for genetic counseling services as the most important one for the field. Increasing coverage of genetic counselors' services is critical to ensuring access because most physicians will not refer patients if insurance companies don't cover the service. Therefore, the Professional Issues Panel will focus on reimbursement strategies from the payer perspective. Members involved with the NSGC's Payer Task Force (see the President's blog of July 23 for more information at http://nsgcpresident.blogspot.com/) will share insights about payers' objectives in regards to genetics services, messages that work to increase utilization and gain coverage of genetic counselors' services, and tools that are or will soon be available to NSGC members for their own conversations with payers.

There are too many wonderful talks planned to touch on all of them, but I know I will be very busy learning from fellow genetic counselors and trying to squeeze in the opportunity to chat informally with many of you as well. I personally look forward to interacting with NSGC members this fall and sharing the highlights of the last year with you. Hope to see you there!

Elizabeth Kearney, MS, CGC, MBA
2010 NSGC President

Tort Reform and Genomics: A Future Odd Couple?

By Sandy Factor, MS, JD

Editors' Note: *Sandy Factor is a genetic counselor who subsequently pursued a law degree (M.S. Sarah Lawrence, Human Genetics, 1978; J.D. Pace, 1988; admitted to the Bar Connecticut, 1988, New York, 1989). She has volunteered to provide a primer to our readership on torts ("civil wrongs") and the intersection of the practice of medical genetics and litigation. This article is the first of two. It provides an overview of medical malpractice litigation, introducing legal terms and definitions. Her follow-up article in the Winter issue of* PGC *will contain case studies highlighting issues in genetic counseling.*

Medical malpractice, product liability for drugs and medical devices, and toxic torts (personal injury lawsuits in which the plaintiff claims that exposure to a chemical caused injury or disease) are pervasive civil litigation matters in both State and Federal courts. This is especially important now that Health Reform legislation has been passed by Congress, and its future financial costs and benefits are of prime interest to legislators.

Using newly evolving technology, genomic medicine will provide products based upon individual genetic risk for diagnosis, prevention, and treatment of both known genetic

conditions and chronic multifactorial population-wide conditions. The manufacturers of these products will also be exposed to litigation, unless specifically "immunized" by legislation. This leaves us asking: How will tort reform affect the current practice of medical genetics? Also, how will the disparate advocacy interests of the corporate, medical, and public health communities influence the future practice of medical genetics/genomics?

Torts – A Background

In general, Tort actions are lawsuits whose purpose is to "make whole," via a monetary award, an injury to a plaintiff ("P"). The defendant ("D") in medical malpractice cases is usually a physician, a hospital and its employees, or a nursing home or, in a products liability case, a drug, chemical, or medical device manufacturer.

The "common law" of torts includes trespass, slander, assault and battery, invasion of privacy, and in a medical context, negligence. In all tort cases, P must allege and prove a loss or harm as the result of an act or failure to act by D (proximate causation). "Fault" by D is conduct that falls below accepted community standards of behavior. It is conduct that creates an unreasonable or unacceptable risk of harm, in violation of a duty imposed by law. The remedy for the harm is "compensatory damages" (money) for both tangible and intangible harm.

The four components of negligence always are: A Duty owed by the D to the P; the Breach of that duty by the D through failure to conform to the required "standard of care"; a sufficient "causal connection" between the D's conduct and the harm; and the actual loss, injury or damage suffered by P.

Medical malpractice is a form of professional negligence. The medical practitioner (such as a physician, nurse, social worker, psychologist, or genetic counselor) ("MP") is alleged to have "failed to exercise the degree of care and skill that is exercised by reasonably well-qualified professionals in that field." The standard of care for MPs is determined by their profession itself; what the other MPs in the same medical specialty ordinarily and customarily do. This is often established by guidance from medical specialty associations.

Informed consent is a significant evidentiary issue in all medical malpractice cases. Tortious conduct may be authorized by P via informed consent. Thus, a harmful outcome will not be actionable if the material risks and alternatives were presented to P. The disclosure must be of information that a "reasonable patient" would want, so as to be able to make an informed choice.

Damages must be proven by P. They always include the actual physical injury suffered bodily, plus any subsequent medical treatments required. This is usually testified to by an expert in the same field of practice as the D. Damages can also include loss of earnings, loss of consortium (spousal intimacy), and pain and suffering (emotional stress). If a P dies as a result of the bodily harm, it is a "wrongful death" as well as a negligence action, and the place of P is taken by P's "estate". In this case, P's heirs benefit from the monetary award.

Proximate causation is the most heavily litigated issue in most medical malpractice cases. Proximate causation means that the departure by the MP from the applicable standard of care caused the injury; it was the MP's sub-standard quality of care that directly caused the harm. In a medical case, the duty of care is established by proof of a physician or other MP-patient relationship. The medical injury is proven by medical records and expert testimony.

Statute of limitations refers to the time limits for a P to initiate a civil lawsuit. Each state has its own legal civil procedures, so the statute of limitations varies from state to state. These time limits protect potential Ds from perpetual exposure to liability claims, allow them and their insurers to "close the books," and prevent evidence from becoming stale and lost over time. A P's time to start the lawsuit begins to "run" when the injury occurs or when it is first discovered. Most states now have a two-year statute of limitations for medical malpractice. It is the responsibility of the P and/or his legal representative to file a timely lawsuit. In addition, the statute of limitations is delayed during the time the P is a minor or mentally incompetent. This time may vary by state from birth to twenty-one years, eighteen years, or ten years. Thus, MPs whose practices deal with fetal, neonatal, or pediatric patients have an extended time frame during which they are at risk of a lawsuit. These MPs should maintain their medical charts and keep their insurance coverage, even after retirement.

In cases where a child P dies after a lawsuit begins, the P's "estate" stands in the place of P. In some states, in cases where an infant patient dies young, allegedly from injuries suffered *in utero*, the estate can bring a "wrongful death" action due to negligence. Some states may also include a stillborn, full-term fetus as a P.

"Wrongful Birth" lawsuits may be brought (not in all states) by parents or guardians if (a) an infant has congenital or genetic defects, (b) the MP should have known before the pregnancy advanced beyond the legal limitation for an elective abortion, and (c) the MP failed to educate the pregnant woman of the potential negative outcome. "Wrongful Life" lawsuits are brought on behalf of a living infant who claims his life is impaired due to a MP's negligence and that the infant would rather not have been born. Very few states permit these lawsuits.

Damage awards for lawsuits by injured infants and children can run into the multiple millions, since severe injuries such as brain damage may require lifelong custodial and medical care. Thus, in some states with a history of large jury awards, Obstetricians (OB) are relocating or retiring from practice, and there are OB residency programs that are not meeting their required "match" with American-trained medical students.

The increasingly high cost of medical liability insurance for physicians, hospitals, other MPs, and their employees has become a significant issue for legislators. The MPs have had escalating yearly premiums for their liability policies, due in part to large monetary awards and frequent lawsuits requiring defense attorneys, even for cases without merit. Insurance is a topic too complex for proper discussion in this article, except to point out that the traditional medical malpractice action, requiring plaintiff and defense attorneys, with expensive discovery, depositions, medical chart review, and expert witnesses testifying in court, may not be the future model for deciding compensation for medical harm.

“Products Liability” litigation is a specialized class of negligence actions. It concerns lawsuits alleging harm from the creation and sale of products of science and technology. The P alleges

harm from use of the product, and must prove that the injury was caused by a deficiency in the way the product was made, labeled or marketed; that the product was "defective" or falsely described. The D tries to prove that the P improperly used the product or that something else caused the injury. Direct-to-consumer genetics test kits are a recent example of a product category that could be at risk for lawsuits, if a P can prove proximate causation and physical injury.

In medical practice, manufacturers of drugs, diagnostic testing kits, surgical implements, and medical devices may be sued, as well as the MPs who use the products for their patients. Such products must be "foreseeably dangerous" (a threshold legal issue) and the manufacturer has a duty to make the product carefully and to provide proper instructions for its use, so as to minimize harm to the patient . The MP has a duty to use the product correctly. For example, there are current reports in the national media about improper use and quality control of radiology equipment for cancer treatment and brain scans, and of improper use of robotic surgical instruments. A hospital has “vicarious liability” for its employees, and a duty to the patient for proper training and supervision of physicians and technicians who operate the equipment or perform tests. A patient has a parallel duty to use the product safely according to directions and instructions, such as taking the proper dosage of a medication at the proper time.

Strict liability for a manufacturer is "liability in tort when an article it places on the market, knowing that it is to be used without inspection for defects, proves to have a defect that causes injury" to the end user. Drugs and medical devices are covered by this standard of care. The manufacturer is under the legal obligation to test its products for safety before selling them, since the consumer has no way of testing for safety.

Pharmaceutical research technology is designed to create "new and better" drugs. United States Food and Drug Administration (FDA)-required human trials are designed to establish safety and efficacy. The future of genomic medicine is based upon the creation, sale and use of such products. Negligence and strict products liability will be an injured plaintiff's mode of recourse if injury occurs.

"Toxic torts" are class action lawsuits which involve multiple injured Ps who allege that they were all harmed by the same harmful/defective product, such as a chemical or drug. Examples include lawsuits against asbestos and benzene manufacturers, tobacco companies, and claims by veterans against the U.S. government for soldiers’ exposure to the pesticide Agent Orange. Hemophiliacs have used class action lawsuits against suppliers of defective blood products, and women have sued silicone breast implant manufacturers alleging a variety of physical harms. These lawsuits are costly for manufacturers to defend and may inhibit the creation and production of potentially beneficial products.

Tort Reform

Legislators in Congress and the states are considering various tort reforms to be enacted into law. The purpose of such legislation is to save (a) taxpayer funds for court costs, (b) corporations, their shareholders, and small businesses the costs associated with medical insurance benefits for their employees, liability insurance, and defense expenses against

lawsuits, and (c) medical providers the high costs of liability insurance premiums. “Tort reform” is a shorthand phrase to signal politicians’ intent to lower the cost of doing business for MPs and manufacturers. Tort reform does not include improvements to consumers’ health care.

Tort reform proposals may include capping compensatory monetary awards to $250,000, removing "pain and suffering" awards, establishing expert panels to weigh evidence, or replacing trials by jury. Congress, however, may not constitutionally be able impose such rules on the states; the issue would ultimately be determined by the Supreme Court. Currently, Ps in products liability cases do "forum shop" for a state jurisdiction with liberal, high-award juries. State legislatures cannot stop individual lawsuits unless they change their residency and "doing business" statutes requirements for initiating a lawsuit.

Conclusion

Genetic and genomic medicine, in the future, will evolve within this complicated legal context. Genomic research may create individualized or “designer” drugs, perhaps to treat one specific individual. Drugs must be safe to be marketed, but how can they be tested? In order to develop a targeted drug, populations of persons sharing a particular ethnicity and, therefore, important parts of their genome, may need to be screened to determine the specific mutation in their genome which raises their genetic risk. Population screening of healthy individuals raises issues of stigmatization, invasion of privacy, and informed consent. These bioethical issues have been dealt with by medical genetics providers for years and are not new. What is new is the desire of government to reduce expenditures for medical care and to reduce litigation for sub-standard care, while at the same time enacting health reform, expanding access to health insurance and medical care. The ethical issues may expand: if the government is paying for care, can they require genetic screening and treatment in high risk populations? If basic public health policy has historically highlighted prevention and education for the greater good for the most people, will the government spend funds for one individual's designer drug? Will genomic medicines be attacked by the same arguments as genetically modified foods?

In summary, the legal issues are complex. This primer provides an overview of the terms and issues, but is not intended as a comprehensive review. The extent of potential future changes under “tort reform” are an unknown at this point. After the November 2010 Congressional elections, the new Congress may take up the issue in committees, if their Chairmen choose to do so. Within their states, legislatures may enact changes to their rules of civil procedure. The follow-up article in the Winter issue of *Perspectives* will provide a practical case study of the liability risks for medical genetics practice under current civil procedures.

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Licensure / Billing & Reimbursement

Coding Corner

By Shanna Gustafson, MS, MPH, CGC and
John Richardson, NSGC Government Relations Director

Coming Soon: An Online Course on Credentialing of Genetic Counselors!

The Coding Corner is supported by the Coding Subcommittee of the NSGC 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 genetic counselors work hard to obtain recognition and reimbursement at our varied institutions, it is clear that many questions still exist regarding how to make this happen. Genetic counselors have been able to obtain their own National Provider Identification number (NPI) since October of 2005 [http://nppes.cms.hhs.gov/NPPES/StaticForward.do]. In 2007, the CPT code 96040 specific for genetic counseling provided by non-physician providers was approved. There are seven states currently issuing licenses to genetic counselors and another six states are writing regulations. However, none of these actions guarantees that any genetic counselor will be professionally recognized or reimbursed as an individual provider by their institution or their regional payors.

It is time that genetic counselors become their own advocates and initiate conversations with their institutions and payors regarding who genetic counselors are, why it is crucial for patient care to ensure that appropriately trained providers provide genetic counseling services, and begin the credentialing and contract negotiations to ensure their services are appropriately reimbursed. While the NSGC continues to work on a national scale to obtain recognition of genetic counselors as health care providers, we need you to reach out locally.

Some of the basic education regarding the complexity of obtaining reimbursement for genetic counseling services was addressed in last years online course, still available at www.nsgc.org, entitled "*Learn the 3 C's to Maximize your Service Delivery Model: Coding, Credentialing and Compliance*." The NSGC is excited to now be preparing a new online course to guide members on how to navigate the process of becoming a credentialed or preferred provider with your institution or payor, entitled "*Take Control of the Revenue You Generate: How to Become a Credentialed Provider."*

"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 Metabolism/Lysosomal Storage Disorder Special Interest Group: A Pro/Con Discussion about Newborn Screening for Lysosomal Storage Disorders

Editors' Note: *As the list of conditions found on newborn screening panels grows worldwide, there is ongoing debate about the inclusion of lysosomal storage diseases (LSDs). In this article, NSGC Metabolism/LSD Special Interest Group Co-Chair* ***Dawn Peck*** *and member* ***Dawn Jacob Laney*** *explain their opposing views about the potential inclusion of LDSs in United States newborn screening programs.*

Newborn Screening for Lysosomal Storage Disorders: A Promise of Hope

By Dawn Peck, MS, CGC

We all, in some ways, fear the unknown. However, without those pioneers who pave the way for future explorers, the unknown may only continue to cause apprehension in one way or another. As a genetic counselor practicing in Missouri, a state that is helping to shape history in regards to newborn screening, I am honored to highlight the positive aspects that will come by further expanding the newborn screening panel to include lysosomal storage disorders (LSDs). In 2009, Missouri made history by being the second state to pass legislation to include a panel of LSDs (Krabbe, Gaucher, Niemann-Pick, Fabry, and Pompe diseases) to our newborn screening panel. Not surprisingly, the legislation for LSD screening was proposed by a family who lost a child to Krabbe disease, in the hopes of providing early diagnosis and treatment options to other families in the state. This expansion forced health care professionals to consider potential future issues while screening for this class of disorders is in its infancy.

Traditionally, we all think of newborn screening as a tool to help identify and treat newborns with potentially life threatening conditions, prior to the onset of symptoms. The proposed panel of LSDs for newborn screening do not all fit the stereotypical threat of "acute neonatal onset" that we typically associate with newborn screening, which forces clinicians out of their traditional comfort zone. The associated morbidity and mortality of classical, early onset disease, availability and feasibility of a screening assay, and new therapeutic options such as stem cell transplant, enzyme replacement and substrate reduction therapy, make it reasonable to consider these disorders for newborn screening. Questions and concerns surface regarding the development of an effective screening strategy (such as confirmatory testing algorithms and follow-up recommendations), and logical treatment recommendations for those identified with late/adult onset or mild disease. There is also the question of treatment and follow-up for atypical phenotypes and pseudodeficiencies, which is already a consideration with some disorders identified by current screening panels (such as 3-methylcrotonyl carboxylase (3-MCC) deficiency, isobutyryl Co-A dehydrogenase (IBDH) deficiency, and short chain acyl-CoA dehydrogenase (SCAD) deficiency). One must embrace the fact that until newborn screening for LSDs is actually underway, the answers to these important questions will continue to remain unanswered.

Prior to screening for phenylketonuria (PKU), affected infants suffered silently for months until they were diagnosed based on the appearance of symptoms. We all know that the damage done at that point would not be reversible. This premise also holds true for LSDs. Infants with Krabbe disease may have the best chance for survival if they are referred for treatment prior to the time that they typically exhibit symptoms (as early as fourteen days of life).

A newborn screening pilot program for Pompe disease in Taiwan demonstrated that early diagnosis by newborn screening impacted the prognosis of identified infants (Chien *et al.*, 2009). In practice, many of the families with children affected with an LSD wish that they knew of the diagnosis earlier, to prevent the 'diagnostic odyssey' that many families experience before the diagnosis is made, provide a prognosis, and offer the opportunity for genetic counseling and the provision of options (such as assisted reproductive technology or prenatal diagnosis) to prevent the birth of a similarly affected sibling. For example, in Gaucher and Fabry diseases, patients may have "nonspecific symptoms" that may result in numerous specialist visits and the provision of numerous possible diagnoses before the correct one is made. Many times, symptoms may appear late enough that a similarly affected child is born by the time the oldest child exhibits symptoms. In addition, another study published in *Pediatrics* (Plass *et al.*, 2010) examined the opinions of Dutch parents and parents-to-be regarding screening for both treatable and untreatable disease. The majority of respondents, especially those with children, were in favor of screening for less treatable or untreatable childhood onset disorders, primarily to prevent the potential of "a long diagnostic quest."

Lessons learned from expanded newborn screening demonstrate that although we may be identifying individuals with variant, atypical, or poorly understood diseases, we must remember the infants identified with classical disease who are benefiting from early diagnosis and treatment. Until screening for LSDs is performed, we will be unable to answer the all important questions regarding effective monitoring and treatment protocols. At this point, I must agree with my colleague, **Dawn Jacob Laney**, (who authored the opposite side of this issue, below) and stress the importance of *multi-disciplinary collaboration* to develop effective follow-up protocols and educational programs in order to make screening for LSDs a success. To quote Missouri's Newborn Screening Laboratory Manager, Patrick Hopkins, the newborn screening community is getting ready to "go where no [wo]man has gone before". In order to be successful in this journey, we must band together and be prepared to face new horizons.

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Newborn Screening for Lysosomal Storage Disease: Proceed with Caution

By Dawn Jacob Laney, MS, CGC, CCRC

In a victory of biochemical persistence, the ability to screen for lysosomal storage diseases (LSDs) in the newborn period has become a reality. As a genetic counselor specializing in the LSDs, this is an exciting accomplishment with the potential to identify at-risk individuals early and begin treatment quickly. However, I have significant concerns that states such as Illinois and Missouri have already legislated a start date to begin selected LSD newborn screening without strong expert treatment recommendations for the newborn period or the implementation of a systemic follow-up plan. Prior to beginning newborn screening for the LSDs, a carefully designed set of treatment recommendations and follow-up protocols for those newborns identified to be at risk must be in place to provide optimal clinical care for patients and their parents.

The proposed newborn screening panel of LSDs currently includes Krabbe, Pompe, Gaucher, Niemann-Pick and Fabry diseases. These conditions were chosen because the screening testing is currently validated for use in the newborn period. Other diseases, such as those in the Mucopolysaccharidosis family, are close to implementation but not yet mandated for a particular state's testing program. Each condition within this panel is very different in terms of onset, treatment options, and disease symptoms; my comments address different aspects of the disease separately.

In clinical care, I have encountered the difficulties in making life-impacting treatment decisions related to Krabbe disease and other infant onset LSDs. Given the morbidity and mortality rates for stem cell transplant, even parents who have a previous child affected with the same LSD can find options, such as pre-symptomatic hematopoietic stem cell transplant in the first month after birth, daunting. As these difficulties occur in families who already are familiar with a particular LSD diagnosis, decisions will be even harder for families without the knowledge of anticipated disease course gained through experience with a sibling. As the reports from New York and their screening for Krabbe disease in the newborn period have already shown in stark detail, each LSD has a very broad spectrum of severity and variants that are not yet fully delineated. The same low enzyme levels on a diagnostic biochemical test may result in severe, childhood onset disease requiring immediate intervention, or result in a later onset phenotype that does not require intervention in the neonatal period. Other than a few individuals who are affected by one of the severe, known, early onset forms linked to specific mutations, it is very difficult to predict clinical phenotype in many LSDs in the newborn period. Given lack of genotype-phenotype correlations and limited knowledge of treatment outcomes, physicians frequently have difficulty determining firm treatment recommendations.

To complicate matters further, many LSDs have exhibited enzyme pseudodeficiencies or molecular variants of uncertain significance that do not result in any clinical phenotype. In order to exclude a pseudodeficiency, treatment decisions may need to be delayed while waiting for molecular testing results. If molecular analysis identifies a variant of uncertain significance or a pseudodeficiency, then the possible disease path is uncertain. Without a strong prediction of clinical phenotype, it is very hard for family members to decide to pursue

a course such as stem cell transplant with significant morbidity or mortality, direct treatment with enzyme replacement therapy alone, or clinical monitoring without treatment.

Along other lines, some conditions (e.g., Gaucher disease or Fabry disease) often have later onset forms that may be difficult to predict and require monitoring and "watchful waiting." As a result of screening in the Ashkenazi Jewish population, many centers are already following asymptomatic children prenatally diagnosed with Gaucher disease without a formalized protocol. In other countries, such as Italy and Taiwan, newborn screening programs for Fabry disease have found an incidence far above our standard reports, as well as subsets of mutations associated with apparently common later onset.

Although the technology is already available, it seems that a series of well-designed pilot programs and professional groups gathered to produce expert newborn screening recommendations, like those created for cystic fibrosis, will help address follow-up and monitoring visits as well as predict a more accurate – real world, if you will – disease incidence. There are already review mechanisms in place, such as the Health Resources and Services Administration/American College of Medical Genetics scientific board that suggests the appropriateness of adding specific disorders to the recommended national newborn screening panel. Although current metabolic centers will handle the clinical load of new LSD diagnoses as states mandate newborn screening for the LSDs, *expert recommendations* to guide physicians unfamiliar with the LSDs will serve to improve uniform clinical care and help patient families make critical treatment decisions.

NSGC News

Testimonials Help Display the Importance of NSGC Branding and the Value of Genetic Counseling

By Amy Sturm, MS, CGC

Most, if not all of us in the genetic counseling profession have been to a social event and asked the question, "So, what does a genetic counselor do?" We answer the question time and time again, but won't it be nice when the day comes that telling a stranger you're a genetic counselor is not met with quizzical eyes? Even within our workplaces, we are asked similar questions, including "What is the value of genetic counseling?"

To help address these issues, the NSGC and SmithBucklin have been hard at work developing and implementing a Brand Platform. The Brand Platform includes all the components necessary to develop a brand, including a brand essence, personality, brand audiences, and a positioning statement and rationale. In order to assist in these endeavors, the NSGC Communications Committee, along with several of the Special Interest Groups (SIGs), have been gathering testimonials from NSGC members to highlight what members think of the branding process. They also have been reaching out to both frequent and newly referring physicians to gather testimonials about the value of genetic counseling services.

Robert Resta provided the following member testimonial:

"In our own minds, the value of genetic counseling is crystal clear. However, we often find ourselves in the frustrating position of (repeatedly) explaining to obstetricians, oncologists, neurologists, internists, and other physicians just what it is we do and why our services are clinically important. With a successful branding campaign, physicians of all stripes will identify genetic counselors with clinical competence, professionalism, and compassion – and feel confident that their patients are in good hands in the complicated world of genomic medicine."

Recent genetic counseling program graduate **Sharanya Kumaravel** stated:

"As a recent graduate, people constantly ask me what I got my degree in. If I were to get a nickel for every person who then proceeded to ask me what genetic counseling was, I probably wouldn't have stressed out about finding a job. The branding efforts of the NSGC give me hope that one day I can tell someone that I am a genetic counselor and they would immediately know my value and role in patient care. As genetic counselors, we already know the importance of our job in the health care setting, but it is time for everyone else, especially physicians and other health care professionals, to also know our importance and to increase collaboration with us in a variety of settings to provide the best patient care."

One of the Co-Chairs of the Cardiovascular SIG, **Christina Rigelsky**, received this physician testimonial from a cardiologist she works with at the Cleveland Clinic Heart and Vascular Institute. Dr. Heather L. Gornik described the benefits she and her patients have reaped by working with a genetic counselor:

"My patients and I have benefited from having a genetic counselor in our vascular medicine clinic. Working with a genetic counselor has allowed us to determine the cause of arterial dissection in many cases. I've found that the counselor can bridge the gap between my ordering a genetic test and making sure a patient receives an appropriate discussion about benefits and risks of undergoing the test and potential implications of genetic testing. The counselors are also a great resource to discuss with and educate patients regarding testing results and have also been very helpful in coordinating testing for family members through resources such as genetic centers in other states."

Elizabeth Hoodfar, genetic counselor and Regional Cancer Genetics Coordinator at Kaiser Permanente Northern California sought a testimonial from an oncologist, Dr. Minggui Pan, who had the following to say:

"Genetic counselors review and confirm a detailed family history and therefore are able to assess risk with comprehensive information, and provide consultation in more meaningful ways and help determine the need of a genetic test for a particular patient.

Genetic counselors do reduce the time we spend with patients on discussing their risk and help us to work more efficiently. I also believe genetic counselors are better trained than most clinicians in assessing a patient's risk and offering further evaluation in more appropriate ways. In this sense, it does reduce liability as well and saves costs since ordering of genetic tests would be by better trained genetic counselors.

Genetic counselors do help patients to understand complex implications of a positive test and what a negative test means as well. It does lead to better outcomes both medically and psychosocially."

These testimonials will help our internal audience understand the importance of the NSGC's branding efforts and will show our external audiences the value of genetic counselors. We hope to highlight these testimonials on the new NSGC Web site and use them in future marketing and branding efforts.

If you would like to provide a member testimonial about NSGC branding, or if you would be willing to gather a testimonial from a physician who uses your services, please contact Meg Orsi at morsi@nsgc.org.

ABGC Update

Thank You to the ABGC Certification Exam Committee!

By the ABGC Board of Directors

With the 2010 exam completed, the American Board of Genetic Counseling (ABGC) Board of Directors is now delighted to have the opportunity to publicly acknowledge the ten members of the Certification Examination Committee (CEC), all of whom worked diligently throughout 2009 and 2010 to develop and finalize this exam.

First, many thanks to **Myra Roche**, Co-Chair of the 2010 CEC. The job of Co-Chair takes much time, persistence, and patience; Myra exhibited all of these and more during her year's tenure as Chair. Myra's attention to detail, along with her focus on excellence and willingness to put in as much time as needed to accomplish the goals for this year, were invaluable to the process. She added new content to this year's Item Writer training program, streamlined the item review process, finalized our new sample exam items and reference list, and rallied our team to develop yet another high-quality exam for 2010.

Next, thank you to our CEC team of ten volunteers, all of whom worked tirelessly to mentor new Item Writers, as well as review and revise items written by all our volunteer Item Writers. The CEC members took personal time – including weekends – to meet at our Test Development office in Kansas, and made themselves available for numerous conference calls to complete the tasks needed to roll out this exam. Every item on the 2010 exam was reviewed and revised on at least four separate occasions, and received final approval by all ten CEC members before being added to the exam.

Thus, we owe a debt of gratitude to this year's Exam Committee members:

Myra Roche (Co-Chair)
Barb Pettersen (Co-Chair/Board Liaison)
Elizabeth Wood Denne
Karin Dent
Shari Baldinger Douglas
Jacky Halliday
Bronson Riley
Jenna Scott
Helga Toriello
Cate Walsh Vockley

Last, but not least, a huge thank you to all thirty of the ABGC and American Board of Medical Genetics diplomates who volunteered to write items for the 2010 exam. Without them, we would not have the quality items necessary to create this exam. Our Item Writers took their task seriously, developing items that both fit the content outline, and reflect the knowledge and skills necessary to practice as a competent genetic counselor.

"It takes a village" to develop our Certification examinations, and we thank our whole village of volunteers for sharing their time and expertise this year.

SIG/Committee Update

Organizational Cultural Competency: The Process of Making the NSGC a More Inclusive Organization

By Patrick Wilson, MMSc, MS, LGC, CGC, Organizational Cultural Competency Working Group Leader for the NSGC Membership Committee
and Sheetal Parmar, MS, CGC, NSGC Membership Committee Chair

The National Society of Genetic Counselors (NSGC) is one of the most forward thinking, cutting-edge professional organizations that anyone could be a part of. Its members are constantly striving to better serve their clients. Genetic counselors are adept at processing a wealth of ever-changing information and presenting it in a manner that is factual, non-offensive, and easily understood by their audiences. However, while the organization continues to maintain the highest level of professional standards in service delivery, it has fallen short in meeting some of the needs of its own members. The NSGC Board of Directors recognizes that in order for the organization and the genetic counseling profession to move forward, the NSGC itself must become more culturally competent.

Cultural competency is the ability to effectively work across cultures. It is not limited to age, race, class, gender, or sexual orientation, but can also include religion, physical ability and other differences. Organizational cultural competency is an evolving process in which an

organization incorporates principles of cultural competence into its practices, policies, and training opportunities to help the organization become more inclusive, respectful and effective to a diverse membership. To the individual NSGC member, organizational cultural competence refers to the development of the ability to communicate effectively and work respectfully with members who are different from them, culturally.

Current and past NSGC leadership have recognized that there is a cultural divide within the organization. Counselors from underrepresented populations and "non-traditional" counseling fields feel excluded. To address this situation, leadership has begun to incorporate culturally competent practices into its structure and the day-to-day workings of the executive office. A diverse group of members with an interest in ensuring that the organization remains relevant to its membership has developed a set of recommendations geared toward strengthening the NSGC in these areas. Currently, a working group within the Membership Committee has been given the task of formally gathering data from the membership as part of a needs assessment.

The Organizational Cultural Competency Working Group is documenting how NSGC members feel about their organization: Does the organization recognize them and what they have to offer to the NSGC and the profession? Is their area of practice respected and encouraged to grow? Information on these and other subjects is needed so that the NSGC can better represent its members and the profession of genetic counseling.

The recommendations from the Organizational Cultural Competency Task Force are currently available on the NSGC website for members to review. The recommendations are divided into five sections: Leadership; Membership Diversity and Training; Data Collection, Public Accountability and Quality Improvement; Integration into Management Systems and Operations; and Community Engagement. Each section focuses on a portion of the organization that needs attention, and leadership is seeking the input of the membership in addressing the concerns presented in the document.

At this year's Annual Education Conference in Dallas, Texas, there will be a focus group meeting. Attendees will have the opportunity to present their concerns and viewpoints with the knowledge that the information will be presented to the organization's leaders. In turn, leadership will use this information to promote an organizational culture that works to respectfully and effectively serve the NSGC's diverse membership.

If you are interested in participating in the organizational cultural competency focus group or would like more information, please contact:

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or
Sheetal Parmar (916-779-3248 or sparmar@pndx.com)

Reference

Olsen L, Bhattacharya J, Scharf A. Cultural Competency: What it is and why it matters. California Tomorrow and Lucille Packard Foundation for Children's Health. 2006.

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Get Involved… Stay Connected! Join the NSGC Mentor Program!

By the NSGC Membership Committee

The NSGC Membership Committee announces the launch of a revamped Mentor Program to begin in November 2010. The program is designed to enhance networking opportunities for NSGC members. Mentors can offer support, guidance, and insight. Mentees can seek advice from peers or learn about a new specialty, among many other benefits of the Program. **NEW** to the NSGC Mentor Program is the ability for mentees to self-match to a mentor through an online matching website. Mentees can choose from a variety of selection criteria to find a mentor who best meets their needs. Discussion topics are also provided on a monthly basis to facilitate continued communication, and a guidebook for mentors will also be available.

The initial Mentor Program pilot project ended in April 2010. The pilot project received many positive reviews and valuable suggestions for improvement from mentors and mentees alike, which have been incorporated into the program. **Katie Dunn**, mentee, commented, "I can't say enough good things about having a mentor. Having a mentor meant having a person cheering for me – a person in my corner. Participating in the mentor program was a valuable experience."

Sign-up for mentors and mentees begins in October 2010. Mentors and mentees of all ages, years of experience and areas of specialty are needed to make the Program a success. The time commitment for participation can be as short as five months or as long as twelve months, and mentors and mentees will decide how often they will contact one another. Look for e-Blasts in the fall announcing enrollment periods.

For those who are still unsure of the benefits of the NSGC Mentor Program, consider mentor **Matthew Tschirgi's** advice: "If you want to expand your horizons and possibly accelerate your career in genetic counseling, get involved in the Mentor Program. You have nothing to lose and much to gain."

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

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Student Forum

"Welcome to Miami, Bienvenido a Miami:" A Multicultural Summer Rotation Experience

By Diana Toledo, Boston University Genetic Counseling Program, Class of 2011

There are many places in the country that offer a diverse patient population, but none may be quite as unique as the population in Miami, Florida. Known as 'The Gateway to the Americas,' Miami has a population of over five million people, 67% of which list Spanish as their first language.[1] The city is diverse, with close to 70% claiming Hispanic or Latino origin (most of which are of Cuban descent), 22% claiming Black, Haitian, or African American background, and fewer than 11% of the population claiming non-Hispanic white background.[2] In deciding where to go for my first clinical rotation during the summer of 2010, I immediately knew I wanted to nose-dive into this multi-cultural Miami population! Needless to say, a summer rotation in Miami provided me with many distinctive experiences that enabled me to learn and grow as a genetic counselor.

I am a Miami native and first American-born daughter to Cuban immigrants. Spanish was my first language, but because I have lived away from Miami for the past six years and have been out of practice, my skills are mostly conversational at this point. I knew that going back to work in my hometown would provide me with the practice I needed to regain my language skills, especially medical terminology and phrases/questions commonly used in a genetic counseling session. I anticipated that the experience of re-acquiring my Spanish language skills would present obstacles, but be well worth it in the end.

My rotation took place at Miami Children's Hospital. During my time there, I participated in and/or observed nearly 100 cases in many different clinical settings, such as general pediatric genetics, craniofacial genetics, Neurofibromatosis, neurogenetics and metabolic genetics, skeletal dysplasias, Tuberous Sclerosis Complex (TSC), and multiple inpatient consults. About half of these sessions were conducted in Spanish as per the patient's request. This served as great experience and practice for me. It felt amazing to be able to communicate with my patients in their preferred language, allowing them to open up and fully understand the information presented throughout the sessions.

Although patients were very receptive to me because I was able to speak with them in their preferred language, healthcare professionals who did not speak the language experienced difficulty building rapport. I followed two medical geneticists during my rotation, one of whom did not speak fluent Spanish. This posed a big problem for her when trying to connect with her Spanish-speaking patient population. One example of this took place in the TSC

[1] "Data Center Results – Miami, Florida" Modern Language Association. 2000 Census. Accessed 8/3/10.

[2] American Community Survey Demographic and Housing Estimates: 2006-2008, Miami city, FL. Accessed 8/3/10.

clinic, where a Spanish-speaking mother presented with her four year-old son who had a clinical diagnosis of TSC. The geneticist spoke to her in English and had me translate for her after she left the office. The patient's mother seemed confused and asked me several questions that I needed to run by the geneticist again. The mother seemed upset about the situation and left the visit not fully understanding the recommended molecular testing, or the reasons that other referrals were made.

I noticed in Miami that many patients have the expectation that all healthcare professionals will be able to communicate with them in Spanish. When that is not the case, patients tend to close off and put up a wall between themselves and the healthcare professional. This gave me insight into how patients who speak a different language perceive the material and me in a counseling session. It reminded me to speak slowly, use shorter phrases, and frequently ask if they need repeating or have any questions for me. In Miami, I observed that the ability to speak the same language as your patients is not seen as a privilege, but a necessity for building good rapport. Conversely, the experiences that I have had in Boston clinics have been quite different. The Spanish-speaking patient population in Boston seems to have more patience with translators and seems to be much more receptive to the session.

Issues that frequently presented themselves during sessions in Miami were those relating to immigration. Many patients I saw during my rotation were recent immigrants from other countries, with most originating from Central and South America. Being a recent immigrant is difficult enough on its own, let alone being a parent of a child with a disability or a genetic condition. They are attempting to learn a new way of life, new language, culture, political and social system, and find employment, as well as dealing with their child's diagnosis or a high-risk pregnancy.

My first case of the summer involved an expectant couple that recently emigrated from Cuba, and discovered that the mother of the baby was a Fragile X premutation carrier. The father of the baby had actually been a doctor in Cuba, but was currently employed as a patient transporter due to the language barrier and the need for him to retake the licensure examination. They were undergoing financial struggles and could not afford to have a child with special needs at this time. They were concerned and anxious about the baby's Fragile X status and opted to have a chorionic villus sampling procedure to find out the results as early as possible in order to make an informed decision about the pregnancy.

Another case in which I participated involved a ten year-old girl who recently immigrated to the United States with her mother from Cuba. The patient had mild intellectual disabilities and had been seen, but not diagnosed, by geneticists in Cuba. A chromosomal microarray was done and she was found to have 22q11 deletion syndrome. I discussed these results with the patient's mother, as well as inheritance and parental studies. She was very grateful that we were able to give her daughter a diagnosis and make recommendations for her daughter's health care, something their country of origin could not do for them. Her father was unfortunately still in Cuba and would not be able to give a parental sample, which is a common occurrence in cases of recent immigrants.

An issue that goes hand-in-hand with immigration is deportation. Not all Miami immigrants are living legally in the U.S. and they risk being deported back to their country of origin. One such case we saw involved a twelve year-old boy with possible Marfan syndrome. The family

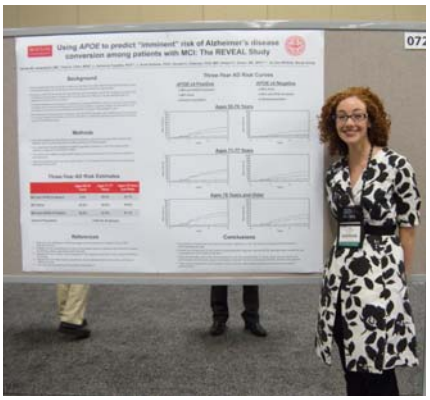
seemed rushed during my history intake, and disclosed to me that they needed to be at an appointment with their immigration lawyer later that afternoon; the boy's father was in the process of being deported to Mexico. The family was clearly distraught and looked to the geneticist I was working with for a letter of medical necessity describing their son's possible condition and the need for his father to stay in Miami. The geneticist was uncomfortable with this request, since she suspected that their son did not have the condition, but did give them copies of the cardiac referral that she was making and helped them as much as she ethically and morally could. The idea of being separated after immigrating to this country together and facing so many struggles would be devastating to a family. This topic occasionally presents itself in clinic in Miami, and was one issue that I learned to manage and deal with in a session.

Deciding to complete my summer rotation in Miami and being immersed in the culture and language after many years of being away was a decision I am very happy I made. I was not only able to hone my skills to complete a medical and family history intake and begin learning my own counseling style, but I had the opportunity to do so in two different languages. Additionally, this experience exposed me to many issues that may arise during clinical encounters, such as language barriers, immigration/deportation issues, and cross-cultural communication. Most of all, I was able to reconnect with my culture, which had long been overdue. It was an amazing learning experience!

The New Graduate Life

Presenting at the 2010 International Conference on Alzheimer's Disease: Stepping out of my Comfort Zone and Traveling to Hawaii

By Denise M. Lautenbach, MS



Using time wisely and perfecting the art of multitasking: these are essential skills for any "type-A" new genetic counselor like myself who constantly seeks balance between being successful at work, building and maintaining personal relationships, and enjoying my mid-twenties. While trying to maintain this balance on a busy schedule, I couldn't think of a better use of my time than reading *A Guide to Genetic Counseling* to prepare for my quickly

approaching American Board of Genetic Counseling Board exam while on a nine-hour flight to Honolulu, Hawaii to present a poster at an international conference. Can you?

It all began back in January, when discussions within our research group about the abstract submission deadline to the 2010 International Conference on Alzheimer's Disease (ICAD) in Honolulu sparked my response of, "I want to go to Hawaii, too!" I am currently the genetic counselor and project manager for the Risk Evaluation and Education for Alzheimer's Disease (REVEAL) Study based at Boston University School of Medicine. Yet, while learning more about Alzheimer's disease and attempting to fit in with non-geneticists is a constant goal of mine, it wasn't quite enough of a motivation for the team to send me to Hawaii; I had to write an abstract and it had to be accepted.

It's strange what the prospect of white sandy beaches, Mai Tais and warm clear water does to a girl. With everything on my plate (i.e., the Boards), the next thing I knew, I had agreed to write an abstract on how we calculated the Alzheimer's disease risk estimates for the upcoming fourth phase of the REVEAL Study, in which we will provide a genetics-based Alzheimer's disease risk estimate to individuals with mild memory problems – a diagnosis termed "mild cognitive impairment" or "MCI." As a new genetic counselor one year out of genetic counseling school, everything has seemed like new territory. Taking this job I had, of course, heard of Alzheimer's disease but had never heard of MCI.

Although my training prepared me to tackle more of the unknowns than the knowns, after I submitted my abstract I experienced some mild anxiety. I realized that I would need to step out of my comfort zone here because, should it be accepted, I would have to present the abstract to people with much different backgrounds than I. Would their questions and concerns be different than mine? Would I be equipped to answer their questions? How will I be perceived as a professional? Would the audience be familiar with genetic counseling and genetic counselors? Would the work be criticized in any way and, if so, will I be able to discuss and defend it? In an attempt to cope with this anxiety, I displaced it by imagining the excitement of traveling to Hawaii and exploring a new state. The abstract was accepted for poster presentation, which relieved some, but not all, of my anxious feelings. I told myself in a pep talk, "Denise, all you have to do is stand by your poster for two hours and talk to people. You know how to have a conversation. You are intimately familiar with this work. You can definitely do this!"

My poster presentation was on the first full day of the conference. I arrived in one of my favorite outfits with an iced Kona coffee in hand, prepared to conquer the day! Once I arrived at the registration desk, I was given a conference bag that was surprisingly heavy. When I opened the bag, I found that there was an entire booklet for each of the six days of this conference. I also saw that there were four hundred posters presented each day. I was absolutely blown away. I had never been a part of something this huge before, making it even more exciting to see my name in the program. As I looked at the first day's schedule, I was again surprised. I didn't realize that there would be full genetics sessions taking place multiple days in a row. Here I thought that I would be the lone genetics person in a sea of neurologists and neuroscientists – boy, was I wrong! As the conference went on, I came to realize that the underlying gist of this conference was more about risk factors, including genetics and biomarkers, and prevention, rather than drug trials, treatments and cures – a taste of the changing climate in how we think of treating neurodegenerative disease. To my surprise, with

my genetics background and my special interest in risk for common disease, the scope of this conference was an excellent fit.

The poster session itself was a rich learning experience for a young professional and researcher. Once I arrived at my poster, there were already people standing near it! I ended up speaking non-stop for two hours, mostly the same conversation over and over again. I met and spoke to a number of interesting and even famous geneticists, one of whom is currently an Editor-in-Chief of *GeneReview*s. To me, this further emphasized how wrong I was in assuming I would be the only genetics person at the conference.

The poster presentation ended, and I marked it as a "success." I still had five conference days left, and as any good multi-tasker would do, I found time to have some fun and enjoy the location of the conference. Sometimes in life, you just have to adopt an "I'll rest and sleep later" attitude. In fact, I found out that you indeed can find time in a single day to attend scientific presentations, snorkel with wild dolphins and sea turtles, eat wonderful food and attend a luau! If only I could be that productive every day.

As intimidated as I was to present research findings at such a large, international conference, I am so glad I had the guts to jump in and do it. A little extra work in preparing the abstract and poster allowed me to have a week away from the regularities of life in a beautiful location. I couldn't help but notice that next year's ICAD conference is in Paris, France. Let the abstract writing continue!

Genetic Counselor Publications

By Jamie Fong, MS

Articles co-authored by genetic counselors
April 2010 – July 2010

(Names of genetic counselors or NSGC members appear in **bold**)

Alter BP, Giri N, Savage SA, **Peters JA**, Loud JT, Leathwood L, **Carr AG**, **Greene MH**, Rosenberg PS. Malignancies and survival patterns in the National Cancer Institute inherited bone marrow failure syndromes cohort study. *Br J Haematol*. 150(2):179-88. 2010.

Bales AM, **Zaleski CA**, McPherson EW. Patient and family experiences and opinions on adding 22q11 deletion syndrome to the newborn screen. *J Genet Couns*. 22 May 2010. [Epub ahead of print.]

Bernhardt BA, **Silver R**, Rushton CH, Micco E, Geller G. What keeps you up at night? Genetics professionals' distressing experiences in patient care. *Genet Med*. 12(5):289-97. 2010.

Causey TN, Bodurtha JN, Ford N. A genetic perspective on infant mortality. *South Med J*. 103(5):440-4. 2010.

Greene MH, Kratz CP, Mai PL, Mueller C, **Peters JA**, Bratslavsky G, Ling A, Choyke PM, Premkumar A, Bracci J, Watkins RJ, McMaster ML, Korde LA. Familial testicular germ cell tumors in adults: 2010 summary of genetic risk factors and clinical phenotype. *Endocr Relat Cancer*. 17(2):R109-21. 2010. [Print Jun 2010.]

McBride KL, **Varga EA**, **Pastore MT**, Prior TW, Manickam K, Atkin JF, Herman GE. Confirmation study of *PTEN* mutations among individuals with autism or developmental delays/mental retardation and macrocephaly. *Autism Res*. 3(3):137-41. 2010.

McGill AK, **Pastore MT**, Herman GE, Alliman S, **Rosenfeld JA**, Weaver DD. A tale of two deletions: A report of two novel 20p13-->pter deletions. *Am J Med Genet A*. 152A(4):1000-7. 2010.

Reis LM, Tyler RC, Schneider A, **Bardakjian T**, Semina EV. Examination of *SOX2* in variable ocular conditions identifies a recurrent deletion in microphthalmia and lack of mutations in other phenotypes. *Mol Vis*. 16:768-73. 2010.

Schilter KF, Schneider A, **Bardakjian T**, Soucy JF, Tyler RC, **Reis LM**, Semina EV. *OTX2* microphthalmia syndrome: Four novel mutations and delineation of a phenotype. *Clin Genet*. 8 May 2010. [Epub ahead of print]

Book review authored by a genetic counselor

(Name of genetic counselor appears in **bold**)

Uhlmann WR. Review of *The Woman Who Walked into the Sea: Huntington's and the Making of a Genetic Disease*, by Alice Wexler (2008). *Am J Hum Genet*. 86(6): 830-1. 2010.

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

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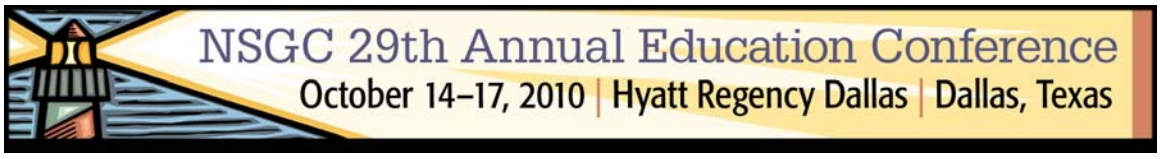
AEC Update

29th NSGC Annual Education Conference



By Shannan DeLany Dixon, MS, 2010 AEC Chair, and
Elizabeth Wood Denne, MS, 2010 AEC Vice-Chair

The Annual Education Conference (AEC) is turning 29! We are thrilled to invite you to join us in Dallas, Texas for the 29th Annual AEC. The Dallas area has so much to offer for attendees. Iconic structures by Frank Lloyd Wright and I. M. Pei, the Dallas Museum of Art, the Morton H. Meyerson Symphony Center, the Campbell Center (home of the Dallas Opera), White Rock Lake Park, the historic Fort Worth Stockyards, the Mesquite Rodeo, the Texas Rangers, the NBA Dallas Mavericks, and the Dallas Cowboys all call the Dallas region home. You should have recently received your preliminary program with all of the dates and deadlines for the AEC, which will be held **October 14-17, 2010**. We look forward to celebrating our 29th year with you.

Pre-Conference Symposia

Are you looking for a more in-depth presentation and discussion about a favorite genetic topic? Don't forget to register for a Pre-Conference Symposium! The Pre-Conference Symposia will take place on **Thursday, October 14**. These optional sessions will offer an opportunity to gain new perspectives and a deeper understanding of six cutting edge topics. Pre-Conference Symposium sessions include:

- 101: Newborn Screening: 2010 and Beyond
- 102: No Boobs Allowed: Update on Cancer Genetic Syndromes and Management for Everything but HBOC
- 103: Genetic Counselors and Clinical Research: A Great Fit
- 104: The Value of Autopsy in the Anomalous Fetus
- 105: Advances in Neuromuscular Disease: Implications for the Field of Genetic Counseling
- 106: Employing Corrective Feedback Techniques in Genetic Counseling Practice

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

Sessions Available Online After the AEC

Once again, sessions from the 2010 AEC will be available online after the conference. In early 2011, a recording of the conference sessions along with synchronized PowerPoint presentations will be available. Register for access to the online sessions before the conference. For those of you who won't be able to join us in Dallas, these recordings will be available for purchase to all members after the AEC and can be used to obtain Continuing Education Units (CEUs).

Program Book: Print Your Notes Before the Meeting

To continue our effort to be "green," speakers' notes and PowerPoint presentations will be posted online prior to the conference instead of including them in the program book. The membership will be notified when the presentations are posted. We encourage you to decide which talks you cannot miss and print out the presentations you want to have on hand at the

conference ahead of time. A suggestion: Read over the presentations on the plane and spend your first night catching up with friends instead of flipping through presentations! The NSGC will offer an internet café, but only for viewing purposes as there will not be printers. If you need to print materials on site, the hotel business center will be available for a fee. If you bring your laptop, another option is to download the presentations ahead of time for viewing during the actual sessions (*Note: wireless Internet is not available in the conference rooms*). A smaller program book will be distributed at registration with the AEC schedule and hotel information, so you will have something in hand to find your way around.

Networking, Networking, Networking

The AEC is not only a great educational opportunity, but a prime networking opportunity. The Welcome Reception is always a good place to see colleagues and friends and should not be missed. Although we have a busy agenda, we are fortunate to offer two networking receptions this year. Please join us for the Welcome Reception on **Thursday, October 14 from 6:00 pm - 8:00 pm**. Additionally, there will be a Networking Reception for all AEC attendees sponsored by the Boulder Abortion Clinic on **Friday, October 15 from 6:00 pm - 7:30 pm**. A list of registered attendees will be posted on the NSGC Web site before the meeting, so you can see who will be at the AEC and arrange networking activities before you arrive.

Outreach Event

The AEC Subcommittee is pleased to announce that we have partnered with Science Teacher Access to Resources at Southwestern (STARS) for our outreach event this year. STARS was developed in 1991 to improve the quality of science education in North Central Texas. At that time, a partnership was formed to make available to middle and high school science teachers some of the vast educational resources of UT Southwestern Medical Center. Since its inception, the STARS Program has grown to serve over 6,000 teachers and 35,000 students in 2,000 schools in the Dallas/Fort Worth area. The scope of STARS has steadily expanded to include over twenty separate programs and projects which are available, free of charge, to teachers and students in the state of Texas (STARS, 2010). NSGC members will be speaking at the STARS October Symposium about the field of genetic counseling and epigenetics. STARS attendees are then invited to attend an Educational Breakout Session at the NSGC AEC on Saturday afternoon. Many thanks to coordinator **Karen Heller** and the AEC Outreach Subcommittee for all of their hard work; we know that this year's outreach event will be a great success.

Abstract Update

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

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

Best Poster Award: As in 2009, a Best Poster Award will be awarded after judging during the "Posters with Authors" session. The winner will be announced later in the conference and will receive a monetary prize.

Late-breaking Session

Based on the success of the late breaking session at the 2009 conference, we will once again have a late-breaking session. However, we have chosen to alter the format for 2010. This year's late breaking session is being planned to address upcoming issues that will impact the entire NSGC community. Details will be included in the on-site Program Book.

Register Now

It's not too late to register for the AEC! Join us in Dallas for a great educational opportunity, while catching up with old friends and meeting new colleagues. For more information, please visit the AEC web page at http://www.nsgc.org/conferences/aec.cfm.

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Elizabeth Wood Denne
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2010 NSGC AEC Vice-Chair

Resources / Book Review

Reviewed by Sophia Ceulemans, MS

A Guide to Genetic Counseling, Second Edition

Edited by: Wendy Uhlmann, Jane Schuette, and Beverly Yashar
Publisher: Wiley-Blackwell
Pages: 624
Retail price: $79.97
ISBN-10: 978-0470179651
ISBN-13: 978-0470179659

The "Purple Book" Turns Green with Envy

After ten years – a lengthy span in the rapidly evolving field of genetics – this premier genetic counseling textbook has been updated (including a refreshing new cover). The Latin adage, *Tempora mutantur nos et mutamus in illis*, comes to mind, which means "Times change, and we change with them."

The eighteen chapters review core competencies, define and demonstrate fundamental counseling techniques, and guide professional development. Each chapter is written by leading experts in the field and contains relevant patient vignettes illustrating key principles. Additional features of the text include concise chapter summaries, concept diagrams, condensed tables, and a list of literature references. Because each chapter emphasizes an isolated component of practice, the "Putting it all Together" case examples at the end are helpful in blending topics and concepts.

Limitations of the first edition included scant guidance about reviewing scholarly articles or conducting original research. An entire chapter on computer-based resources quickly became outdated. Some readers desired more practical information regarding counseling techniques, such as suggestions for defining genetic concepts to clients. Many of these first edition criticisms have been addressed in the new version. An added chapter on genetic counseling research outlines the process of designing, implementing, and analyzing research data, including ethical considerations. An entire chapter is devoted to risk communication and decision-making, an area supported by an expanding body of literature that barely existed when the first edition was published.

Some of the updates from the first edition reflect a change in technology or even how we view our profession. The authors realized that citing Web addresses is not particularly useful in today's world of ephemeral hyperlinks, so the chapter on computer-based resources was eliminated. This left room for coverage of more current topics, such as genetic counselors as educators and the expanding opportunities for genetic counselors. The NSGC's 2010 Professional Status Survey highlighted a growing number of genetic counselors in non-clinical positions, and the second edition of this text has attempted to incorporate this shift.

This book remains one of the best reference materials of the field, but areas for improvement remain. Study questions at the end of each chapter – for readers to consider individually or as a small group – would help integrate key principles and build vocabulary around genetic counseling and its complexity. These questions or points to ponder could also guide professionals pursuing certification. While this is, after all, a textbook, some of the chapters are dense and heavy with citations. These chapters may benefit from simple examples or a sentence that points the interested reader to a more comprehensive source. Additionally, the inclusion of chapter numbers in the page headings would further enhance readability.

Finally, some of the patient vignettes that included multiple counseling visits for the same indication seem impractical and unrealistic for most busy clinic settings. Many genetic counselors have altered their practice of counseling to incorporate briefer encounters or have explored alternative service delivery models, like telemedicine or group counseling. These alternative models require creative use of fundamental counseling skills, and vignettes illustrating these approaches may be helpful.

Undoubtedly, there will be times when the field of genetic counseling undergoes an identity shift, perhaps initiated by changing technology and improved testing platforms, a growing understanding of genomic health, or differences in genetic service delivery models. Nevertheless, the ideas grounded in this text will help shape the profession during these defining times.

Like the first edition, this text is designed to supplement genetic counseling coursework and clinical exposure to facilitate skill development. The intended audience is current students or practicing professionals in the field of genetic counseling, although other healthcare professionals with an interest in the field may also benefit from this resource.

Media Watch

By Claire Noll, MS, CGC

(Names of genetic counselors appear in **bold**)

Editors' Note: *Space limitations preclude listing items available on websites aimed at patient education. Priority is given to interviews or mentions in traditional and electronic media. All links are active at the time of editing. Subsequent to editing, some archived links may have expired. We apologize in advance if this occurs. Thank you to all the genetic counselors who have submitted items for Media Watch, and please keep them coming!*

May 11, 2010 – *United Press International* (www.upi.com/Health_News/2010/05/11/Genetic-tests-need-genetic-counselors/UPI-30351273624958/)
"Genetic tests need genetic counselors"
In connection with the announcement that Walgreens would sell genetic test kits, the United Press International picked up the NSGC press release in which **Elizabeth Kearney** discussed genetic testing. She recommended that "people should first meet with a genetic counselor to determine whether genetic testing is right for them and to prepare them for what they might learn."

May 12, 2010 – *Minneapolis-St. Paul Star Tribune*
(www.startribune.com/lifestyle/health/93606064.html?page=1&c=y)
"DIY genetics put to test"
This article appeared after the Walgreens announcement but before its retraction, and opened with a futurist's prediction that "[i]n five years, every newborn will leave the hospital with his or her complete genetic code." **Matt Bower** cautioned that "it's really unclear how useful this stuff is… There is no pill or treatment that will prevent Parkinson's disease or slow it down."

May 14, 2010 – *NBC's The Today Show* (www.msnbc.msn.com/id/37119557/ns/health-health_care/)
"Walgreens gene test on hold after FDA alert"
Following the reversal of the Walgreens decision to market genetic test kits, The Today Show aired a segment in which the kits were discussed by their health expert, who pointed out that the test results provide information that may be of little utility. **Ellen Matloff** agreed that uninformed test users could interpret the results incorrectly, with reactions such as "I guess I shouldn't have children" or "I'm going to die at a young age – let me spend all my money and drop out of college."

May 14, 2010 – *Dallas Morning News* (www.dallasnews.com/sharedcontent/dws/news/nationworld/stories/051410dnmetgenetickit.3f00046.html)
"Over-the-counter DNA kit worries genetics experts"
Another article about the Walgreens retraction clarified the concern of some experts that the markers used in the kit were poorly understood or insufficiently actionable. "We're very concerned that this general information will be misinterpreted," said **Linda Robinson**. "Not all genetic tests are equal, and the public doesn't realize that."

May 30, 2010 - *MSNBC* (www.msnbc.msn.com/id/37407874/ns/health-health_care/) "Gene tests have answers, but do we want them?" and *Live Science* (www.livescience.com/health/personal-gene-tests-disease-risk-100530.html)
"Scrutiny of personal gene tests increases"
In the context of the Walgreens news, **Barbara Bernhardt** was interviewed about a personalized medicine research project on which she collaborates. Participants in the study are offered the chance to have genetic testing; if they accept, they must undergo genetic counseling first. She disclosed that early results indicated that participants appeared to understand the basic genetics very well, that about one-third of them shared test results with a physician, and that many participants stated they would make lifestyle or health changes but typically had already intended to do so before receiving genetic results.

June 1, 2010 – *Las Vegas Review-Journal* (www.lvrj.com/health/genetic-testing-a-mixed-blessing-95300664.html)
"Family history: Genetic testing a mixed blessing"
This article also explored genetic testing in the context of the Walgreens news, though with reference to the genetic services available in the local area. **Erica Ramos** pointed out that more people are aware of genetic counseling now. "For certain patients, it's part of their standard medical care," she said.

June 9, 2010 – *KOCE Real Orange* (http://video.koce.org/video/1549296955/)
"Healthy Orange segment on hereditary cancer"
Jeanne Homer clarified the differences between sporadic and hereditary cancer and encouraged people interested in cancer genetic counseling to obtain as much family history information as possible. She emphasized that gender-specific cancers can be inherited from either parent, and discussed the protection offered by the Genetic Information Non-Discrimination Act legislation.

June 17, 2010 - *CNN Health* (www.cnn.com/2010/HEALTH/06/17/home.genetic.tests/index.html?eref=rss_health&utm_source=feedburner&utm_medium=feed&utm_campaign=Feed%3A+rss%2Fcnn_health+%28RSS%3A+Health%29)
"Should you test your genes?"
A pediatrician described her decision to order genetic testing using a direct-to-consumer (DTC) kit when it became available for a reduced price and despite the doubts raised by the Walgreens decision. For others who think they might want DTC testing, some questions to consider about the goals of testing were provided. "At the very least, before you do online testing, you should identify a genetic counselor in your area who could help you interpret the results," said **Ellen Matloff**.

June 28, 2010 – *Scientific American* (www.scientificamerican.com/article.cfm?id=personal-genome-sequencing)
"Genome sequencing for the rest of us"
This article compared the predictions made following the successful sequencing of the human genome ten years ago with today's reality of far fewer medical applications, particularly for complex diseases, than hoped for. "We naively thought that there would be a few genes involved in risks for a variety of common disorders," said **Barbara Bernhardt**. With regard to direct-to-consumer genetic tests, she added, "They certainly do provide medical information to people. From that perspective, they do need to be regulated as medical devices."

July 1, 2010 – *Jewish United Fund* (www.juf.org/news/local.aspx?id=61266)
"Genetic testing requires more than a saliva sample kit"
Another article prompted by the Walgreens news offered a cautionary note with regard to direct-to-consumer testing because this type of analysis is based on new technology and new science. **Michele Gilats** used the example of a currently available kit that provides a risk for Parkinson's disease based on a single marker. "You may read that you have a greatly increased risk for Parkinson's, but you also need to be aware that this disorder has less than one percent basis in genetics. The problem is, it's the initial risk number that's going to stick in people's minds," she said.

July 12, 2010 – *Winnipeg Free Press* (www.winnipegfreepress.com/life/health/know-your-pedigree-family-medical-history-holds-key-to-your-health-98248729.html)
"Know your pedigree: family medical history holds key to your health"
This article reminded readers that despite all the attention on genetic testing, family history remains an important part of genetic risk assessment. **Cheryl Shuman** stated that learning about cancer from relatives may be difficult when cancer is considered a private subject. **Rochelle Demsky** added that in such cases, it may be difficult to learn the correct cancer diagnosis (such as cervical versus ovarian cancer) because the older generation did not ask as many questions of their doctors. **Karin Dent** mentioned that despite these possible problems, she is seeing more and more of her patients coming to consults having already obtained a lot of family medical history. "In fact, they seem to go out of their way to collect it before they see us, because they recognize that it's something that can help us help them," she said.

July 13, 2010 – *Anderson Independent Mail* (www.independentmail.com/news/2010/jul/13/mobile-genetics-lab-takes-science-lessons-students/)
"Mobile genetics lab takes science lessons to students"
The Greenwood Genetic Center, with funding from the National Human Genome Research Institute, has outfitted a bus with a fully functional science laboratory in order to bring genetics activities to area teachers and students. "We're also able to discuss the ethics of testing and introduce students to careers in genetics," said **Lori Bassett**.

July 16, 2010 – *Business Wire* (www.businesswire.com/portal/site/home/permalink/?ndmViewId=news_view&newsId=20100716005941&newsLang=en)

"Informed Medical Decisions named URAC award finalist for best practices in health care consumer empowerment and protection"
Informed Medical Decisions, Inc. was named one of thirty finalists for a health care industry award that will be presented in October 2010. **Kelle Steenblock** described the main benefit of the company's business model as "eliminating essentially every barrier to delivery of genetic counseling services" through cost-effectiveness and convenience.

July 20, 2010 – The State – South Carolina's Homepage (www.thestate.com/2010/07/20/1382717/live-longer-by-talking-about-health.html)
"Live longer by talking about health at family reunions" and
July 23, 2010 – Fort Worth Star-Telegram (www.star-telegram.com/2010/07/23/2357088/family-reunions-are-healthy-in.html#tvg)
"Family reunions are healthy in more ways than one"
As the suggestion to share family medical history at family reunions takes root, genetic counselors are seeing the consequences. "I've had examples in my practice where a family member across the country had a genetic test done, found they had an inherited trait, and contacted a family member here," said **Karen Brooks**. "Then they call me and say they want to come in and be tested for this trait."

August 2, 2010 – *ScienceRoll* (http://scienceroll.com/2010/08/02/national-society-of-genetic-counselors-interview-with-elizabeth-kearney/)
"National Society of Genetic Counselors – Interview with Elizabeth Kearney"
A self-described "genetic blogger" who clearly supports genetic counseling in association with direct-to-consumer testing interviewed **Elizabeth Kearney** on that subject. "Seeking genetic counseling prior to genetic testing is an excellent way to protect against potential harm to consumers who do not have much knowledge or experience with genetics. Sometimes tests will provide helpful information and sometimes the information is either not helpful or even alarming," she explained.

Summer 2010 – *Cure Today*
(http://www.curetoday.com/index.cfm/fuseaction/article.show/id/2/article_id/1483)
"The genes that bind"
This article in a magazine for families touched by cancer addressed the question of when to discuss risks and testing with the children of a parent who carries a mutation in a hereditary cancer gene. **Amie Blanco** described the reluctance of some families to talk about Lynch syndrome as their children grow older, and the need to be blunt if they postpone the talk for too long. "There always seems to be, in this young adult group, some life transition that they are going through that parents are using as a reason for why they should hold off," she said. In families with a *BRCA* mutation, **Beth Peshkin** suggested giving frank but limited information to pre-teens. She advised, "You don't always have to give the comprehensive answer. You can answer the question that the child has without going into more than he or she wants to know necessarily."

Please submit Media Watch items to Claire Noll at genecounselor@gmail.com

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Research Network

By Emily Place, MS, CGC

Genetic Basis of Inherited Reproductive Disorders

The Reproductive Endocrine Unit at Massachusetts General Hospital is conducting a research study to learn about the hereditary basis of reproductive disorders. Individuals with precocious puberty, delayed puberty, or absent puberty (i.e. Kallmann syndrome, hypogonadotrophic hypogonadism), and individuals with adult-onset disorders (i.e., hypothalamic amenorrhea, very low testosterone) are eligible. Participation involves a blood sample, family history collection, a questionnaire, and olfactory testing. Screening is conducted for abnormalities in *KAL1*, *FGFR1*, and a growing list of candidate genes. Results may be released to participants, if desired.

Contact: Margaret Au, MBE, MS, CGC at 617-726-5526 or ReproEndoGenetics@partners.org

MOMS – The Management of Myelomeningocele Study

MOMS – The Management of Myelomeningocele Study is actively recruiting pregnant women for a randomized clinical trial designed to compare prenatal surgery versus standard postnatal surgery for spina bifida. Screening begins by telephone and a review of medical records. Interested candidates who qualify are assigned to one of the following three MOMS Centers for a comprehensive evaluation: The Children's Hospital of Philadelphia, the Vanderbilt University Medical Center in Nashville, or the University of California San Francisco. Eligible candidates who decide to participate are randomized to the prenatal surgery group or the postnatal surgery group. Participants must complete enrollment by 25 weeks gestation.

Contact: Jessica Ratay, MS, CGC at 1-866-275-6667 or moms@bsc.gwu.edu; www.spinabifidamoms.com

Measuring internalized stigma in first-degree family members of someone with a severe mental illness

Do you have a parent, child or sibling with schizophrenia, schizoaffective disorder, or bipolar disorder? Researchers at the University of British Columbia have developed a questionnaire to measure stigma in first-degree relatives of individuals with a severe mental illness. The study is enrolling parents, siblings, and children of people with schizophrenia, schizoaffective disorder, or bipolar disorder to complete a two part study questionnaire. No travel is required. Interested individuals can participate by telephone or mail/email.

Contact: Emily Morris, Research Coordinator, at (604) 875-2000 ext. 4732 or mental.illness@ubc.ca

DNA Biobank and Cell Repository

The National Institute for General Medical Science (NIGMS) supports a DNA biobank and cell repository at the Coriell Institute for Medical Research in Camden, New Jersey. This biobank supplies scientists worldwide with materials facilitating research on the diagnosis, treatment and prevention of disease. The NIGMS is currently collecting samples from individuals with inherited genetic diseases and chromosomal abnormalities. A limited number of samples will be accepted for each disease, so please contact us to determine eligibility for sample acceptance. Interested donors may contact Coriell to receive a blood collection kit. Donation requires completion of patient/parental informed consent, submission form, and a clinical information summary. Coriell covers the cost of shipment.

Contact: Tara J. Schmidlen, MS, CGC at (856)757-4822 or tschmidl@coriell.org; http://ccr.coriell.org/

The Simons Variation in Individuals Project

The Simons Variation in Individuals Project (VIP) is characterizing individuals with 16p11.2 deletions and duplications. Participants must have a documented 16p11.2 deletion or duplication. Both biological parents must participate and be willing to travel for a minimum of two days at one of the study sites which include Baylor University, Emory University, Children's Hospital of Boston, and the University of Washington. Travel expenses and accommodations will be paid. The visit will include a medical, neurological, and psychometric assessment and MRI. Research findings will be shared with the families, and a web-based community for 16p11.2 families will be developed.

Contact: Andrew Faucett, MS, CGC at 404-778-8420; www.simonsvipconnect.org

Genetic Contributions to Autism Spectrum Disorders

The Program in Genomics at Children's Hospital Boston is conducting a study to identify genes that influence the development of autism spectrum disorders (ASD), as well as identify gene expression profiles that could be used as diagnostic and prognostic tools for ASD. Children older than eighteen months of age with a diagnosis of an ASD are eligible to participate. Medical and family history information will be obtained from participants via telephone or mail. Additionally, a blood or saliva sample is required. Blood draws can be performed by a participant's physician. There is no cost to participate and travel to the Children's Hospital Boston is not required.

Contact: Caitlin Kreitman, BS, Study Coordinator, at 617-919-3490 or Caitlin.kreitman@childrens.harvard.edu

Williams Syndrome

We are looking for adults with Williams syndrome to take part in a study called Genetic Analysis of Human Disorders at University of Connecticut Health Center, Department of Reconstructive Sciences (University of Connecticut Health Center IRB Number: 09-199-1). Participation would involve a one-time visit that would last up to 45 minutes. This visit would include discussing the study and obtaining informed consent, as well as a tiny skin biopsy from the upper arm after the doctor applies a numbing medication. A skin biopsy can also be collected during any surgical or dental procedures planned in the near future. Participants will receive a $50.00 check by certified mail for their time and inconvenience.

***Contact**:* Ginger Nichols, MS, CGC, at (860) 523-6458 or GNichols@uchc.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* gene replacement therapy trials for LCA appear promising, a molecular diagnosis for LCA is increasingly needed.

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

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