Perspectives in Genetic Counseling

Volume 29, Number 1
Spring 2007

Jessica Mandell, MS
Editor
jmandell@slc.edu

President's Beat

Who Wouldn't Want to be a Genetic Counselor?

There may be some people who would not want to be a genetic counselor, but I am not one of them! For me this has been an incredibly rewarding profession. Every time I hear about a genetic counselor forging a new trail into some unknown territory, I get excited about how much we can do with our degree. I realize not everyone feels the way I do. I know there is dissatisfaction with salaries and opportunities for advancement, but hopefully, with the hard work and dedication of all of us, this will keep changing in the positive direction.

A Troubling Trend
Given my own enthusiasm for the profession, it is hard for me to understand the recent trends in our graduate programs. We were fortunate that for many years the pool of applicants had grown; however, in the last four years the number of applicants plateaued and has started to slightly decline. Is this a trend in all graduate programs or health professional programs, or is it specific to genetic counseling?

As I reflect on our strategic initiatives and professional organization, we need to think critically about the longevity of our profession. If we do not continue to have young people interested in the profession, who are we doing this for, and who will continue our efforts in advocacy, industry, public health, research and client care? We do not want to be in the same situation as our other genetics colleagues, faced with dwindling numbers. But how do we continue to entice and encourage young people to be genetic counselors and other genetic professionals as well?

Talk Us Up
One of our most powerful assets is *you*, the members of NSGC. Maintain your willingness to discuss your profession, to allow those interested to observe cases, to offer internships, to speak to students in grade school, high school or college. Even the person sitting next to you on the airplane (although you must be prepared for them to put on their earphones…) offers an opportunity to spark interest in genetics professions. Think back to how you first became interested in genetic counseling; you may owe it all to one of your colleagues.

Grad School True and False
It is important to know the facts about admission into graduate programs and trends in our profession. The climate has changed over the past 10-15 years. What was true when you applied might not be true anymore. For instance:

Myth: It's almost impossible to get into a genetic counseling program.
Fact: While about 36 percent of those applying to a genetic counseling program are accepted, of those who interview, closer to 57 percent are accepted.

Myth: If you don't get into a genetic counseling program, don't bother trying again.
Fact: If a candidate has strengthened their application, a second application is welcome and often successful. (Please encourage candidates to call program directors to get feedback on what they can do to strengthen their application!)

Myth: Applying to as many programs as possible maximizes the chances of getting accepted.
Fact: While it's true that applying to more than one program is a good strategy, most applicants apply to an average of five programs.

Myth: Programs are so hard that many people are not able to complete their degree.
Fact: Only about one percent of those accepted to a program do not finish, for a variety of reasons.

Myth: A large number of genetic counselors are leaving the field to pursue other professions.
Fact: Few genetic counselors have left the field to pursue a career that is completely unrelated to their professional degree.

Myth: Genetic counselors are leaving the field because of limited opportunities for professional growth.
Fact: Nearly 90 percent of graduates in the past 10-12 years are working in the genetic counseling field, either clinically or in an expanded role using skills acquired with their degree.

If every member of NSGC could influence just one person, we would have 2,300 new people interested in a profession in genetics. You have the power to help secure the future of the genetic counseling profession. Go forth; be true to your mission, and good luck.

Cathy Wicklund, MS
NSGC President

Data provided from the Association of Genetic Counseling Program Directors, 2006.

Career Watch

ACOG Practice Bulletin Number 77: What does it mean for the practice of genetic counseling?

By Karen Heller, MS

In January 2007, the American College of Obstetricians and Gynecologists (ACOG) published its *Practice Bulletin Number 77: Screening for Fetal Chromosomal Abnormalities.*[1] The Bulletin redefines obstetricians' practices for offering prenatal screening and diagnostic testing to their patients, and the implications for genetic counselors are profound.

Historically, the top two indications for genetic counseling referral have been "advanced maternal age" (AMA) and "positive maternal serum screen." This new Practice Bulletin effectively eliminates both of these categories of patients.

No More AMA, No More Positive Screen

According to the Bulletin, maternal age should no longer be used as the criterion for offering prenatal diagnostic testing. The Bulletin states, "screening *and* invasive diagnostic testing for aneuploidy should be available to all women who present for prenatal care before 20 weeks of gestation regardless of maternal age" (emphasis added). Furthermore, it is suggested that screening results no longer be reported as either positive or negative using an "arbitrary cutoff." Rather, "it is preferable to provide patients with their numerical risk determined by the screening test…" so that the patient can make her own decision about diagnostic testing.

In this new paradigm, ALL pregnant women are to be explained the various options of screening and invasive diagnostic testing. Some may choose diagnostic testing without screening. Those who choose screening would make a subsequent decision about diagnostic testing based on their "individual risk assessment," rather than an arbitrary determination of whether the screen is positive or negative. It also is acknowledged that some patients may decline both screening and testing.

GCs Left Out?

Where does this leave genetic counselors? Obstetricians are expected to discuss with patients the difference between screening and diagnostic testing at the outset. Some patients may select the option of proceeding directly with diagnostic testing and thus may see a genetic counselor on their way to the perinatologist. But what about the majority of patients selecting screening first? Their results will not be labeled as "positive" or "negative." It is not feasible for a genetic counselor to see every patient after screening to explain the significance of her "individual risk assessment" and help her decide whether to proceed with diagnostic testing. Once again, the burden falls on the obstetricians, and the genetic counseling session is relegated to simply obtaining "informed consent" for the diagnostic procedure if it has been selected. The only mention of genetic counseling in the Bulletin is, "Some patients may benefit from a more extensive discussion with a genetics professional or a maternal-fetal medicine specialist, especially if there is a family history of a chromosome abnormality, genetic disorder or congenital malformation."

Additional Controversial Topics

The Bulletin addresses other concerns about what types of screening to offer patients. Practitioners have been debating the relative merits of first trimester screening utilizing free beta-hCG versus total beta-hCG. Exclusive patent holders of the former assay have maintained its superior performance. Data from the FASTER trial, however, confirm that both are effective,[2] and the Bulletin refers to the two analytes as interchangeable.

Another polarization has centered on preferences for first trimester versus second trimester versus integrated screening. The latter option is further complicated by the possibility of "contingent" or "sequential" approaches. ACOG advises that women be offered first trimester or integrated screening if they have access to NT (nuchal translucency) certified sonographers and CVS. If these requirements are not met, serum integrated screening is "ideal" for women presenting in the first trimester.

Obstetricians are legitimately concerned about the overwhelming number of screening methods available, and the Bulletin agrees, stating that, "It is not practical to have patients choose from among the large array of screening strategies...". Genetic counselors can

partner with obstetricians to evaluate their patient population and the available community resources to help decide which screens to offer.

Redefining our Role

As genetic counselors, we need to determine how our services are best utilized in the proposed new model of obstetric practice. Perhaps we become more involved in general education of obstetric patients at the primary care level, somehow assisting primary care providers in explaining different screening and testing options and interpreting risks. Although referrals for positive family history may increase, elimination of the categories of "AMA" and "positive screen" will surely reduce referrals for genetic counseling.

Furthermore, with growing evidence that the risks of both amniocentesis and CVS are lower than what we have traditionally told patients[3,4], diagnostic testing may become more routine, and obstetricians may be less inclined to refer those patients for genetic counseling. Unless we are proactive in reevaluating our role, demonstrating our value for the obstetricians and their patients and creating an acceptable referral indication list, we stand to lose some of the strong partnerships we have forged with our obstetrician colleagues.

References

1. Screening for fetal chromosomal abnormalities. ACOG Practice Bulletin No. 77. American College of Obstetricians and Gynecologists. *Obstet Gynecol*. 109:217-227. 2007.

2. Canick JA et al. Comparison of serum markers in first-trimester Down syndrome screening. *Obstet Gynecol*. 108:1192-1199. 2006.

3. Eddleman KA et al. Pregnancy loss rates after midtrimester amniocentesis. *Obstet Gynecol*. 108:1067-1072. 2006.

4. Caughey AB et al. Chorionic villus sampling compared with amniocentesis and the difference in the rate of pregnancy loss. *Obstet Gynecol*. 108:612-616. 2006.

Note from the NSGC President

Thank you to Karen Heller for submitting her personal and professional insights about the new ACOG guidelines. As our prenatal colleagues are well aware, ACOG practice bulletins can and do have a substantial impact on the practice of genetic counseling. At our recent meeting, the NSGC Board of Directors reviewed the guidelines and was pleased to note that genetic counseling is an integral part of the recommendations. However, it is up to us to continue to define the gold standard of genetic counseling content and delivery. NSGC has reached out to the ACOG leadership regarding ways that NSGC members can collaborate with ACOG Fellows in facilitating the implementation of the clinical management guidelines at the local level. This may include many of the areas outlined in the above article, including professional education of and outreach to our primary care colleagues regarding the role of referral to trained genetic counselors in the screening and follow-up protocols.

Cathy Wicklund, MS
NSGC President

Special Series: Cases in Expanded Metabolic Screening

In 2005, the American College of Medical Genetics' Newborn Screening Expert Group recommended a uniform panel of 29 conditions for which state newborn screening programs should test newborns.[1] *These recommendations not only expand the number of diseases on the test, they require prenatal and pediatric genetic counselors alike to determine the impact and recurrence risk of unfamiliar metabolic conditions. In response to the expanded panel, the Metabolism/Lysosomal Storage Disease Special Interest Group is presenting a four-part series in* Perspectives *on several lesser-known genetic conditions that are now included in newborn screening. These cases are intended to help both metabolic and non-metabolic genetic counselors as they come face-to-face with these "unusual" diseases.*

Case 1: 2-methylbutyryl-CoA dehydrogenase deficiency (2-MBADD)

By Amy White, MS and Dawn Jacob Laney, MS

Disease Review

Biochemistry: Deficiency of 2-methylbutyryl acyl-CoA dehydrogenase deficiency, an enzyme involved in the breakdown of isoleucine; 2-MBADD is classified as an organic acidemia.

Genetics: Autosomal recessive; ACADSB gene located at 10q25-q26; Common mutation in Hmong population: M356V.

Incidence: 1 in 500 individuals of Hmong ancestry (an Asian ethnic group from mountainous regions of southern China); Rare in other populations, although three asymptomatic infants of Northern European ancestry have been detected by newborn screening in Wisconsin.

Natural History: This condition was first reported to cause severe neurologic decompensation, hypoglycemia and hypotonia in four children from two families, one of European and another of Pakistani descent. Recently, a symptomatic child from Somalia has been reported. Two symptomatic individuals of Hmong descent in Wisconsin have been found to date, showing developmental delays that resolved after initiation of a low protein diet, carnitine supplementation and avoidance of fasting. Over 40 Hmong infants identified by newborn screening in Wisconsin have been asymptomatic.

Genetic Counseling - Positive Newborn Screening in a Hmong Infant

A one month-old male is referred to metabolic genetics clinic in Wisconsin following an abnormal newborn screen result that showed an elevation of C5 isovaleryl-carnitine. His parents are recent Hmong immigrants in their mid-thirties. A Hmong interpreter is present for the visit. The parents are told that, most likely, their child has 2-methylbutyryl-CoA dehydrogenase deficiency (2-MBADD), a common genetic condition in the Hmong population that results from a problem with protein metabolism. It is explained that a few Hmong

[1] Watson, MS, et al., "Newborn screening: Toward a uniform screening panel and system - Executive summary," *Pediatrics*, Vol. 117 No. 5 May 2006, pp. S296-S307

infants with this condition have shown developmental delays in the first two years of life that resolved with low protein diet, carnitine supplementation and frequent feedings. The majority of Hmong infants with 2-MBADD detected by newborn screening, however, are completely asymptomatic and do not require a special diet or medication.

The parents are counseled regarding limiting protein intake in infants with 2-MBADD to the recommended daily allowance and providing carnitine supplementation for the first year of life. These are protective measures, since there is no way to know which children may become symptomatic.

Discussion of inheritance proves difficult, as the interpreter helps translate a “common language” for genetics. The parents are told that 2-MBADD is passed down through the blood of one generation to the next and results from a combination of blood from the mother and father. The pedigree reveals four older siblings, none with developmental delays. Testing is offered to the parents and siblings at no cost. The parents decline testing but agree to treatment for their infant. A low protein recipe for formula and a low protein powder supplement are provided. The parents also receive a booklet on 2-MBADD in Hmong and English.

At a three-month follow-up visit, the parents state that they have not used the low protein formula or powder supplement. Their infant son is growing and appears healthy, and they do not believe he has any medical problem.

Teaching Lessons

1) Limited Knowledge of Natural History
It is difficult to provide genetic counseling for 2-MBADD in the Hmong population because the natural history of this condition is still being determined. At this time, the majority of individuals identified by newborn screening are asymptomatic. Even with attempts to treat all infants detected by newborn screening in the first year of life, at least 50 percent of families in Wisconsin are not compliant with treatment when they report back during follow-up visits. To date, none of these children have shown developmental delays. The metabolic genetics professionals in Madison and Milwaukee are working on a study to clarify the natural history of this condition in the Hmong population.

2) Language/Cultural Barriers
As with all languages, there are specific nuances to Hmong languages. First, there are several dialects, all of which were originally limited to oral transmission with no written form. Today, White Mountain Hmong has developed a written form that is the most widespread dialect. An education group developed the parent booklet written in both White Mountain Hmong and English that was given to the parents in this case (see picture).

Second, Hmong languages do not have equivalent words to explain inheritance such as sperm, egg, genes, chromosomes, etc. Therefore, this condition is described merely as being passed down through generations by blood, which conveys the right meaning when translated. In addition, the word “chemical” should be avoided when explaining biochemistry, as it is often assumed to mean an unnatural compound similar to a poison. The English word “substance” is more acceptable and more easily translated.

3) Lack of Compliance
There are several reasons for noncompliance with treatment for 2-MBADD in the Hmong population. Hmong parents feel that a child who appears healthy does not have a medical problem. When interacting with medical professionals, Hmong individuals are likely to agree

as a sign of respect, even if they do not believe what they are being told or do not intend to follow the recommendations. Most importantly, parents think they are receiving mixed messages by being asked to bring their child into clinic for a medical problem when almost all children with this condition are not sick.

Publications by Genetic Counselors

By Deborah McDermot, MS

This is the second installment of a new feature in Perspectives *highlighting the publication activities of genetic counselors in peer-reviewed journals (other than the* JOGC*). Each issue will list the articles published during the previous quarter and will spotlight one publication in which a genetic counselor served as first or senior author. Featured papers are chosen at the discretion of the* PGC *Editorial staff.*

Featured Paper

(Names of genetic counselors appear in bold.)

McWalter KM, Hughes CM, Masakawa NK, Minatoya KR, Miyake S, Oyadomari TA, Shimamoto JH, Parlin L. Community recommendations on outreach activities for QUEST-expanded: Medicaid managed care for the aged, blind and disabled population. *Hawaii Medical Journal*. 66:36-40. 2007.

Kirsty McWalter is a genetic counselor with the Hawaii Department of Health, a long way from her home in British Columbia. She credits her supervisor, **Sylvia Au,** with the state's strong genetics component which employs three additional genetic counselors. Kirsty devotes about half of her time to public health functions, including clinical research, grant writing and education at venues including universities, high schools, career fairs and physicians' offices. The other half of Kirsty's position involves clinical genetics activities, primarily pediatrics. In Hawaii, these clinical services are collaborative and funded largely through the Department of Health, the University of Hawaii Medical School and local hospitals to service the island of Oahu, neighboring islands and soon an outreach clinic to Guam.

Outreach Education

Kirsty became involved in a regional LEND (Leadership Education in Neurodevelopmental Disabilities) program, as a community participant advising on genetics. This opportunity proved to be quite informative for her, as she became intimately aware of how hard families touched by genetic conditions and other issues had to fight for the services that they need. Her involvement also educated the other participants in the LEND program unfamiliar with genetics or genetic counselors. Her featured publication is an extension of her work in LEND.

Managed Care Concerns

Kirsty's biggest challenge with this community-based study was the focus on qualitative analysis. Community focus groups illuminated that parents and caregivers for those with genetic conditions felt that genetic services must be protected within the state's proposed shift to a managed care Medicaid model. While the publication does not pertain to a purely genetic topic, it highlights the multiple roles that genetic counselors are taking in a changing

medical environment and the powerful influence such participation can have on our professional colleagues. The findings from this study were presented formally to the Hawaii State Legislature in December 2005, and Kirsty and her colleagues are awaiting the final decision on how clinical genetic and other services may or may not be affected.

Power in Numbers

Kirsty's involvement in LEND and the community outreach project have led to an increase in referrals for genetics consults from a number of other professionals in the program, which she predicts she would not have received prior to taking part in these committee functions. "We all want the same things for our patients, and there is power in numbers," she said about these new professional relationships.

Articles Co-Authored by Genetic Counselors, Dec 2006-Feb 2007

Austin JC, Smith GN, Honer WG. The genomic era and perceptions of psychotic disorders: Genetic risk estimation, associations with reproductive decisions and views about predictive testing. *Am J Med Genet B Neuropsychiatr Genet*. 141:926-8. 2006.

Chang L, **Blain D,** Bertuzzi S, Brooks BP. Uveal coloboma: Clinical and basic science update. *Curr Opin Ophthalmol*. 17:447-70. 2006.

Clark HM, Gamm J, Huether CA, Buncher CR, **Pfau RIB, Warren NS.** Genetic counselors and research: Current practices and future directions. *Am J Med Genet Part C: Semin Med Genet.* 142:276-283. 2006.**

Grady, MC. Preconception and the young cancer survivor. *Matern Child Health J*. 10 (Supp. 7):165-168. 2006.

Jayadev S, **Michelson S,** Lipe H, Bird T. Cambodian founder effect for spinocerebellar ataxia type 3 (Machado–Joseph disease). *J Neurol Sci*. 250:110-3. 2006.

Kushner JD, Nauman D, Burgess D, Ludwigsen S, Parks SB, Pantely G, **Burkett E,** Hershberger RE. Clinical characteristics of 304 kindreds evaluated for familial dilated cardiomyopathy. *J Card Fail.* 12:422-9. 2006.

Li D, Parks SB, **Kushner JD,** Nauman D, Burgess D, Ludwigsen S, Partain J, Nixon RR, Allen CN, Irwin RP, Jakobs PM, Litt M, Hershberger RE. Mutations of presenilin genes in dilated cardiomyopathy and heart failure. *Am J Hum Genet.* 79:1030-9. 2006.

Lipinski S, Lipinski M, Biesecker LG, **Biesecker B.** Uncertainty and perceived personal control among parents of children with rare chromosome conditions: The role of genetic counseling. *Am J Med Genet C: Semin Med Genet.*142:232-240. 2006.**

Madeo AC, Pryor SP, Brewer C, Zalewski C, King K, Butman JA, Yang Y, Park H-J, Griffith AJ. Pendred Syndrome. *Semin Hear.* 27:160-170. 2006.

Norris VW, Arnos KS, Hanks WD, Xia X, Nance WE, Pandya A. Does Universal Newborn Hearing Screening identify all children with *GJB2* (connexin 26) deafness? Penetrance of GJB2 deafness. *Ear Hear.* 27:732-741. 2006

Ricker CN, Hiyama S, Fuentes S, Feldman N, Kumar V, Uman GC, **Nedelcu R, Blazer KR,** Macdonald DJ, Weitzel JN. Beliefs and interest in cancer risk in an underserved Latino cohort. *Prev Med*. Oct 5, 2006. [Epub]

Ruttum MS, **Reis LM,** Semina EV. Application of genetic approaches to ocular disease. *Pediatr Clin North Am.* 53:751-65. 2006

Shannon KM, Smutko GC, Niendorf KB, Ryan PD. Uptake of BRCA1 rearrangement panel testing in individuals previously tested for BRCA1/2 mutations. *Genet Med.* 8:740-745. 2006.

Sutton E, Rosapep L, **Truit M,** Ball K, **Biesecker B,** Guidotti R, McLean D. Through the viewfinder: Positive exposure a year later. *Am J Med Genet C: Semin Med Genet.* 142:260-268. 2006.**

******* *Find more publications by genetic counselors in a special issue of AJMG Part C: Seminars in Medical Genetics, vol 142(4), November 15, 2006. Issue title: "Toward Evidence-Based Genetic Counseling," edited by* ***Bonnie Baty*** *and* ***Barbara Biesecker.***

NSGC News

Get to Know the NSGC Executive Office

To better acquaint NSGC members with the Executive Office Staff, Perspectives *is featuring profiles of those involved in the behind-the-scenes operations of NSGC. This issue features* ***Meghan Carey,*** *Operations Manager****.***

What are your primary responsibilities for NSGC?
I am the administrative/operations manager for NSGC. I help **Kristen Smith,** the NSGC Executive Director, oversee the general membership operations. I also coordinate the activities of the other staff who work on NSGC in marketing, education and programs, conference and tradeshow services and accounting.

What is your background experience?
I have worked for healthcare associations for seven years and have worked in non-profits since 1995. I have a Master's in Social Work with a concentration in non-profit administration.

What do you find interesting about NSGC?
I really enjoy getting to know the NSGC members and learning about the wide variety of settings in which they work. I am fascinated with the field of genetic counseling, and I love learning about all of the issues that NSGC is involved with. The energy that NSGC members have for this organization and for your profession is really amazing and that makes it fun to work with you.

What are your hobbies?
Outside of work I enjoy running and hiking. I am a member of a book club that has been meeting for over six years. I also love visiting my friends who are now scattered all over the country.

AEC Update

"New Discoveries Daily"

By Karen Powell, MS and Courtney Rowell, MS, 2007 AEC Co-Chairs

We are looking forward to seeing you at the 26th NSGC Annual Education Conference (AEC) in Kansas City, Missouri – where you will find "New Discoveries Daily." In May you will receive your program brochure in your mailboxes with all of the dates and deadlines for the 2007 AEC. **Remember, the AEC is held early this year, October 13-16. Therefore, important dates for the AEC are coming up soon.**

- Abstracts for platform or poster presentations are accepted from **now through April 30, 5:00 pm EST.** See the article below on abstract submission for more details.

- The deadline for early registration for the short course and AEC is **July 20.**

- The short course, entitled "Survival Skills for the 21st Century: How to Shape Your Future as a Genetic Counselor," is scheduled for **October 11-12.** This course covers the evolving roles of genetic counselors and the variety of skills we need in order to develop genetic counseling programs and our careers.

- This year we are bringing you an advanced program called, "Emerging Technologies in Genetic Teaching and Learning." This electronic extravaganza is scheduled on **Friday, October 12,** between the short course and the AEC. Whether you're a technological novice or a savvy high tech user, this program is sure to please! You will be able to test-drive many tools, from "wiki's" to "videogoogle" to "voiceover PowerPoint."

- The AEC includes two outreach events this fall. An educational event with local Kansas City high school students takes place on **Friday, October 12.** NSGC also is partnering with the Genetic Alliance to invite eight leaders of advocacy organizations to attend the AEC and network with NSGC members. Watch for more information about these exciting opportunities.

- We are staying at the Westin Crown Center, located in the Hallmark Crown Center area of Kansas City. Westin's are known for their "heavenly" beds. Be sure to make your reservations early – the hotel will hold rooms until **October 9.**

The 26th NSGC AEC promises something for everyone, from fetal surgery to the role of genetic counselors during mass disasters to ways of enhancing your upward mobility in our profession. We hope that you plan on joining us in Kansas City!

Contact: Karen Powell, klpowel2@uncg.edu
Courtney Rowell, courtney.rowell@yahoo.com

Call for Abstracts – Platform or Poster Presentations

The 2007 AEC Abstract Committee invites the submission of abstracts to be considered for platform or poster presentation at the 26th NSGC AEC, October 13-16 in Kansas City. Submissions of abstracts of interest to the genetic counseling profession and related fields

are being accepted now through April 30 (5:00 p.m. EST). Authors will be notified of the review committee's decisions in June. Submissions from full members, students and non-members are encouraged. Monetary awards will be presented for best full member and student member abstracts.

Please carefully review the instructions for submission of abstracts; on the NSGC Web site (www.nsgc.org) click on the "Conferences" tab and choose "Abstracts" from the drop-down menu, or go directly to www.nsgc.org/conferences/aec.cfm.

Contact: NSGC Executive Office, nsgc@nsgc.org or
Jehannine Austin, MS, jcaustin@interchange.ubs.ca, or Sara Cooper, MS, sccoope@emory.edu, AEC Abstract Committee Co-Chairs

---

Student Column

Grab Your Boards for the Summer

By Stephanie Herbert, MS

The lazy days of summer are quickly approaching. Sun, surf, vacations, picnics. Everyone's favorite time of… oh, what did you say? No summer this year? That's right… it's Board Exam time! But before you decide to put that sunscreen completely away, you may want to hear about this.

A New Review
A new concept in Board exam review has been created to make this summer a little less hectic. The University of Pittsburgh is sponsoring the 9th Review Course in Medical Genetics & Genetic Counseling. The course is being managed by BLCommunications, under the direction of **Bea Leopold** as managing director.

This year's course is unique because it will be offered exclusively online. The new format is "created to meet the study and pacing preferences of test takers, to save travel-related expenses and logistics for our registrants and to provide continuing education units for practicing genetic counselors who need to fulfill their CEU requirements but are unable to travel."

Register Now
Registration for this online course is now open. Course topics include cytogenetics, molecular cytogenetics, biochemical genetics, cancer genetics, malformations and dysmorphology, quantitative genetics, molecular genetics, prenatal clinical genetics, screening, counseling and ethics – and more.

Visit www.blcommunications.us for more information and registration materials. Group discounts are available.

Five Great Reasons to Register for the University of Pittsburgh Online Review Course in Medical Genetics and Genetic Counseling

1. Study for boards when and where you want. You can even download the lectures to your iPod or MP3 player.

2. Brush up on medical genetics and earn up to 3.0 CEUs toward recertification, right from the comfort of your desktop, or at the gym.

3. Save hundreds of dollars in travel expenses; do the math and think of the possibilities!

4. Download study materials in PowerPoint including outlines, tutorials with instant feedback, pre-tests and post-tests. Bonus! You can email questions to specific faculty members.

5. Start studying ASAP – 30 hours of audio-downloadable lectures go live in March.

The University of Pittsburgh Department of Human Genetics Genetic Counseling Program gratefully acknowledges support of this program provided by NSGC.

Book Review

A Different Kind of Perfect: Writings by Parents on Raising a Child with Special Needs

Editors: Cindy Dowling, Neil Nicoll and Bernadette Thomas
Publisher: Trumpeter Books, Boston, 2006, 245 pages, $22.95
Reviewed by: Danna Loder, Genetic Counseling Student, McGill University

A Different Kind of Perfect is a collection of real life stories written by parents of children with disabilities. It aims to provide invaluable insight into the lives of families affected by disabilities – from the challenges and frustrations to the joys and triumphs. The short stories offer comfort and support to parents and provide understanding to family, friends and professionals.

Focus on Emotions

The stories are organized into chapters, each representing the different emotions parents may experience with a child with disabilities. The chapters address denial, anger, depression, acceptance, empowerment, marriage, family and friends, love and joy, spirituality and laughter. **Neil Nicoll,** a family and child psychologist specializing in developmental disorders, introduces each chapter with a definition of the emotion. He explains its purpose, its positive or negative effects and methods of coping. The introductions are scientifically accurate but not overly technical. Each story then gives a compelling account of how and why the emotion arose and the effects it had for the families involved.

A Personal Objective
Editors **Bernadette Thomas** and **Cindy Dowling,** both parents of children with disabilities, were unsatisfied by the available selection of parent support books. Such books were "…overtly sentimental and painted parents as selfless martyrs who accept their fate stoically." A *Different Kind of Perfect* is straightforward and honest. The stories reveal parents' true feelings, some of which may be viewed by society as inappropriate or may not be often discussed. For example, some parents reveal their negative feelings toward their children and their immense difficulty in losing their dreams and expectations. Others discuss their endless tears, fears and frustrations. At the same time, stories in which parents have overcome these negative emotions and learned to cope with everyday challenges and to embrace life exemplify the strength and hope that can be achieved when raising a child with disabilities.

Diverse and Similar
The families in the book represent a wide variety of ethnicities, socioeconomic classes, religious/spiritual beliefs and family structures. Despite these differences, all parents of children with disabilities can relate to these stories, learning that they are not alone and that life with a child with disabilities can be incredibly rewarding. Professionals, family and friends also can gain important insight into what life is like raising a child with disabilities. Such understanding can lead to increased empathy, support and advocacy.

Public Eye

Media Watch

By Roxanne Ruzicka Maas, MS

October 6, 2006 – *South Reno Weekly*, "Translating Genomic Science Into Clinical Practice"
Robbin Palmer wrote this article for her local newspaper. She also was a guest on "High Desert Forum," a call-in radio program on local public radio station KUNR, where she discussed hereditary breast cancer.

November 4, 2006 – ScienceDaily.com, "New Study Finds Amniocentesis Safer For Pregnant Women"
This article commented on the findings of the FASTER trial, reported in the November issue of *Obstetrics & Gynecology*. The study indicated that pregnancy miscarriage rates after routine mid-trimester amniocentesis are significantly lower (1/1600) than rates previously published (1/200). In reference to amniocentesis, the article claimed that "most women fear them while doctors recommend them."

December 5, 2006 – *The New York Times*, "Wanting Babies Like Themselves, Some Parents Choose Genetic Defects"
A study to be published in *Fertility and Sterility* explores how some parents intentionally choose "malfunctioning" genes for their offspring, producing disabilities like deafness or dwarfism. An estimated three percent of American prenatal genetic diagnosis (PGD) clinics have used PGD "to select an embryo for the presence of a disability" because "it turns out that some mothers and fathers don't view certain genetic conditions as disabilities but as a way to enter into a rich, shared culture." Unlike the online magazine *Slate,* that reportedly

called this methodology "the deliberate crippling of children," *The New York Times* article acknowledged that, "traditionally, cultures were perpetuated through assortative mating, with intermarriage among the like-minded and the like-appearing." As such, "…the use of PGD to select for deafness may be merely another ritual to ensure that one's children carry on a cultural bloodline."

December 11, 2006 – *MSNBC Newsweek*, "Peering Into the Future"
This article explored how genetic testing is transforming medicine and the way families think about their health. Advances in testing bring difficult choices but also new abilities to ward off disease. Examples included PGD for Huntington disease, genetic testing for hereditary cancer and amniocentesis to detect isovaleric academia in a child so that neonatal treatment could save her from severe disabilities. **Wendy Uhlmann** is quoted regarding the questionable value of genetic testing for a condition when treatment is not available. The burgeoning field of pharmacogenetics and the business side of genetic testing were mentioned.

December 12, 2006 – *The Washington Post*, "Teaching Hospitals How to Listen"
After being diagnosed with cancer and struggling for attentive care during treatment, a woman relayed her difficult experiences convincing administrators that staff responsiveness – or lack of it – affects patient outcome. She noticed parallels between her work as a psychologist with autistic children and the morale of hospital patients depending on the social contact they receive by medical staff. By voicing her concerns about the long wait she endured at one hospital and the lack of responsiveness by staff at another hospital, she affected change in the first hospital's policies of scheduling patients and documenting wait times and received a written response from the second hospital's ombudsman.

January 14, 2007 – WashingtonPost.com, "What's Lost in Prenatal Testing"
The mother of a child with Down syndrome expressed her concern about the ACOG "recommendation" that all pregnant women "get" prenatal screening for Down syndrome. She was most concerned that increased prenatal screening would lead to more terminations of pregnancies with abnormalities and stated, "There is a fundamental societal misperception that the lives of people with intellectual disabilities have no value." She quoted The Hastings Center finding that 80 percent of genetics professionals would terminate a pregnancy with Down syndrome and commented that "these are the people advising pregnant women." She hoped physicians and genetics professionals could learn from people with disabilities and their families and disseminate the message that all people have value and dignity.

January 29, 2007 – *MSNBC.com Newsweek Society*, "Golly, What Did Jon Do?"
Like the WashingtonPost.com article, this was another individual's reaction to the ACOG statement that all pregnant women should be offered prenatal screening. The author described prenatal screening as "a search-and-destroy mission," stating that "diagnosing Down syndrome can have only the purpose of enabling – and, in a clinically neutral way, of encouraging – parents to choose to reject people [with Down syndrome] as unworthy of life."

Licensure Update

NSGC Forges Ahead on Legislative Action Plan

By The Billing & Reimbursement Task Force

Since NSGC's multi-tiered strategy for achieving licensure and improved billing and reimbursement for genetic counseling services was unveiled at the Annual Education Conference in November, Billing and Reimbursement Task Force representatives have been involved in a flurry of activity to enact central elements of the plan. Stay tuned for updates via E-blasts and NSGC Web site postings during March and April for detailed information on the intricacies of the federal and state legislative endeavors and the importance of licensure.

Future issues of *Perspectives* will summarize key initiatives, provide an overview of guiding principles for bills along with model legislation and update members about progress on licensure efforts in various states. In addition, those attending the 2007 Region I, II and IV meetings will have an opportunity to ask questions about the billing and reimbursement strategic plan from various members of the NSGC leadership. For a summary of NSGC's strategic plan, please read the Winter 2006 issue of *Perspectives*.

Spring Cycle of State Licensure Grants Call for Applications

The spring cycle of the State Licensure Grants is now open for applications. Proposals are due to the NSGC Executive office by Friday, April 13. Send proposals via email to nsgc@nsgc.org and to sgoldman@dhs.ca.gov. The email subject line should read "Licensure Grant Award."

States that have drafted bill language should include this text in the proposal so it can be reviewed by the Licensure Subcommittee. Licensure grant funding will be available by June 1 and must be spent by December 31. Full details of the application process and Licensure Grant Awards are available on the NSGC Web site at www.nsgc.org/members_only/licensure/Licensure_Grant.cfm.

Contact: Sara Goldman, MS, 510.412.1463, sgoldman@dhs.ca.gov

SIG Update

News from the Familial Cancer Risk Counseling SIG

ASCO Slides Now Online

The Familial Cancer Risk Counseling SIG is pleased to announce the availability of the American Society of Cancer Oncology (ASCO) curriculum, "Cancer Genetics and Cancer Predisposition Testing, 2nd Edition," on the NSGC Web site. The slide presentation is available for download and is posted in two areas of the Members Only section:

1) "Useful Tools for Cancer Genetics Counselors" in the Familial Cancer Risk Counseling link under SIGs

2) the cancer section of Virtual Slide Swap under "Tools for Your Practice."

The complete ASCO CD may be requested by contacting the NSGC Executive Office at nsgc@nsgc.org. We hope you take advantage of this useful resource.

Founding Members of the Genetic Counseling Foundation
The Familial Cancer Risk Counseling SIG has been granted the honor of Founding Member status by the Genetic Counseling Foundation (GCF). The 2006 SIG members made a generous donation in support of the newly established GCF. Great achievements through the GCF are sure to come thanks to the involvement of caring groups and individuals such as the 2006 Familial Cancer Risk Counseling SIG members. Thanks and congratulations!

Professional Status Survey in Cancer Genetics
The Familial Cancer Risk Counseling SIG currently is preparing the "Professional Status Survey 2006: Cancer Genetics Analysis." This report highlights employment experience, patient volume, salary, benefits and job satisfaction of counselors spending 50 percent or more of their time in cancer genetics. This report will be available soon. There will be an E-blast announcement to SIG members, and the report will be posted on the NSGC Web site via the Members Only section in SIGs/Familial Cancer Risk Counseling.

Contact: Nancie Petrucelli, MS, petrucel@karmanos.org, or Joy Larsen Haidle, MS, joy.larsen.haidle@northmemorial.com, Familial Cancer Risk SIG Chairs

---

Research Network

Genetic Identification of Cardiac Conditions

The Molecular Cardiology Laboratory of the Weill Medical College of Cornell University is enrolling individuals with a variety of cardiac-related conditions in ongoing gene identification projects. Specific conditions of interest include: Familial Aortic Aneurysm, Familial Congenital Heart Defects, Noonan syndrome, Carney complex and cardiac myxomas and Trisumus pseudocamptodactyly.

Contact: Deborah A. McDermott, MS, 212.746.2054, dam2001@med.cornell.edu

Hematological Malignancies in Families

The Hereditary Cancer Institute at Creighton University is enrolling individuals from families with two or more hematological malignancies. The study is investigating genes related to the following familial cancers: multiple myeloma, ALL, AML, CLL, CML, Hodgkin disease and non-Hodgkin disease. Any combination of the above hematological malignancies within a family is of interest.

Contact: Kelly Ferrara, MS, 800.648.8133, kellyferrara@creighton.edu

High Resolution Marker Chromosome Analysis at No Charge to Patients

Emory University is offering molecular cytogenetic analysis of marker chromosomes for prenatal and postnatal samples (with the exception of the common satellited markers derived from chromosome 15). Researchers will determine the origin and euchromatic content of the marker using a high density microarray, in conjunction with a well characterized set of pericentromeric FISH probes. In marker chromosomes that contain euchromatin, the size of the imbalance and the gene content will be determined for prognostic purposes.

This clinical investigation is part of an NIH study conducted by **Drs. David Ledbetter** and **Christa Lese Martin** in the Department of Human Genetics at Emory University. There is **no cost** to participants. The laboratory is CLIA certified, allowing for results to be released.

Contact: 404.727.7098, research@molecular-rulers.org, www.molecular-rulers.org

Multiple Studies Ongoing at Mount Sinai

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

Contact: Kerri Lee, MS, 212.241.6012, kerri.lee@mssm.edu

Novel Genetic Risk Factors Contributing to Breast Cancer

Washington University School of Medicine is recruiting women nationwide to participate in a study on the genetic risk factors for breast cancer. Women diagnosed with invasive breast cancer before 40 years of age are invited to participate. Women who have undergone genetic testing of the BRCA1, BRCA2, p53, pTEN, e-cadherin or other genes are eligible, regardless of the test results. Women who have not had testing also are eligible.

Participants are required to provide cancer-related medical records and family history and submit a blood sample. Parents of participants are invited to join as controls.
No travel is required. Study materials and shipping supplies for samples are sent to participants by mail. Blood samples can be drawn locally. There is no expense to the woman, her parents or her physician.

Contact: Jennifer Ivanovich, MS and Paul Goodfellow, PhD, Principal Investigators, 314.454.5076, jen@ccadmin.wustl.edu

Resources

Featured on www.nsgc.org – NSGC Speakers Bureau

NSGC is pleased to announce the new and improved Speakers Bureau. The Speakers Bureau has been developed as a resource for individuals and groups at all levels seeking NSGC members to speak on topics related to genetics and the genetic counseling profession. Whatever type, size and location of speaking engagements you're interested in, there's a spot on the Speakers Bureau for you! If you've signed up in the past, please take this opportunity to view and update your record. If you've never signed up, please enroll yourself today.

Visit the Speakers Bureau at www.nsgc.org/speaker_bureau/. Follow the link to add yourself to this resource (or to edit your record). You will be prompted to enter your NSGC Web site log-in and password.

Both the public and NSGC members will be able to search for speakers. Speakers can be identified locally or nationally or by presentation topic. If you are in need of someone to speak on issues of importance to genetic counselors at your next event, visit the NSGC Speakers Bureau today.

Contact: NSGC Executive Office, 312.321.6834, nsgc@nsgc.org

Bulletin Board

Call for Nominations

NSGC Board of Directors

The nomination process is open for the NSGC Board of Directors positions of President-Elect and Regional Representatives for Region II and Region V. Regional terms are for three years and the Presidential term is for four years, starting January 1, 2008. The Call for Nominations process closes May 18. For more information or to submit a nomination, please visit www.nsgc.org/members_only/index.cfm.

ABGC Board of Directors

The American Board of Genetic Counseling (ABGC) is seeking genetic counselors who have been Board certified for five or more years, to serve on the ABGC Board of Directors.

Nominations are being accepted through Monday, April 23, after which a slate of four candidates will be chosen by the Nominating Committee. Elections will take place in August 2007 for two positions whose five-year terms will begin January 1, 2008.

- To make a nomination, send the name of a potential candidate to Beth Balkite, MS, ABGC Executive Director, xdabgc@nc.rr.com.
- For questions regarding serving on the ABGC Board of Directors, contact LuAnn Weik, MS, lweik@chw.org, or visit www.abgc.net.

2007 ABGC Nominating Committee: Richard Dineen, Chair, LuAnn Weik, Board Representative, Mary Ahrens, Barbara Harrison and Daniel Riconda

NSGC Awards: Deadlines Approaching

The Audrey Heimler Special Projects Award

The deadline for the 2007 Audrey Heimler Special Projects Award is May 15. In the past, awardees of the $5,000 grant have used funds for projects such as creating educational materials for patients, developing practice guidelines and exploring ways to involve new genetic counselors in the NSGC leadership structure. For more information or to submit an application, please visit www.nsgc.org/members_only/funding/ahspa.cfm.

The Jane Engelberg Memorial Fellowship

This prestigious fellowship, awarded by the NSGC, is in memory of **Jane Engelberg,** MS (Sarah Lawrence College, 1973). A bilingual genetic counselor, Jane developed expertise in counseling patients on issues related to hemophilia and prenatal diagnosis during her 15 year career in New York City. Jane died of Hodgkin's disease in 1988. The JEMF was established in 1990 by Jane's husband, **Alfred B. Engelberg,** and is funded by the Engelberg Foundation.

The JEMF Award strives to promote the professional development of individual counselors and to improve the practice of genetic counseling by providing support for scholarly investigation of any aspect of the profession.

A one-year, $50,000 award is granted to an individual genetic counselor or counselors. Applicants must be genetic counselors with Board certification (ABMG or ABGC) or active-candidate status (ABGC) and who are full members in good standing with NSGC.

Applications are available at www.nsgc.org. Follow links to "Members' Corner" and "Funding Opportunities." Applications are due by Monday, May 7. **Please note our IRB policy has changed. Applicants must obtain IRB approval upon notice of funding but not at the time of submission!**

Contact: Betsy Gettig, MS, Chair, JEMF Advisory Group, 412.624.3066, betsy.gettig@hgen.pitt.edu

.

JEMF Advisory Group: Betsy Gettig, Chair, Michelle Fox, Secretary, Kathy Valverde, Financial Chair, Toni Pollin and Jill Stopfer

Letters to the Editor

A Sad Day for the Genetic Counseling Profession

Dear Editor:

In late 1985, 37 years after the inception of the American Society of Human Genetics (ASHG) and six years after the incorporation of the NSGC, **Dr. Beverly Rollnick** made history as the first genetic counselor elected to the ASHG Board of Directors. This was a momentous event and a major milestone for the genetic counseling profession. The ASHG is the oldest organization in the medical genetics establishment and was the sole professional home for all who had chosen careers in human genetics until 1980.[1] I still recall the excitement of finding two members to sponsor my application to ASHG, receiving my acceptance letter and attending my first ASHG meeting. Though still in school, I felt like I clearly had arrived.

Within 1987, other genetic counselors were elected to the ASHG Board of Directors and the profession benefited from nearly two decades of continuous representation. This fall, **Bonnie Leroy** was nominated for a three-year term that would have begun January 1, 2007. Her qualifications are impeccable—years of national leadership and service along with academic accomplishments in research, teaching, scholarship and publication that would make any ASHG member proud. But Bonnie did not win this election. The reason? The number of genetic counselors who belong to ASHG (both the absolute number and the proportion of the membership) has been diminishing annually.[2] At the end of 2006, genetic counselors made up only four percent of the ASHG membership, with the future looking bleak for us to regain our seat at this table of leaders.

Is this what the genetic counseling profession wants? Is this lack of representation truly in our best interest?

All human genetics professionals, regardless of their degree or focus, are trained in the tradition of a multi-disciplinary team such that the whole is enriched by the sum of its parts. Yet genetic counselors are not participating in either the ASHG, where we are full voting members and can serve on committees as well as be elected to the Board of Directors, or the American College of Medical Genetics (ACMG),[3] where we can sit on all committees, network with medical genetics colleagues and participate in decision-making related to genetic services. Why is this happening?

National involvement in genetic organizations is about more than paying dues and getting a journal. It is about coming to the table, broadening one's perspective, interacting with a range of colleagues and making contributions. It is about the solidarity that is derived from having a voice. Above all else, participation and representation of genetic counselors in our sister genetics organizations should be an equally shared responsibility and an integral part of being a genetics professional. Losing a genetic counselor on the ASHG Board of Directors must serve as a call to action for the entire genetic counseling workforce. Membership information is below. Act now – we deserve to do better!

Judith Benkendorf, MS
Bethesda, MD

For membership information and application please go to the following links:

ASHG: www.ashg.org/genetics/ashg/membship/001.shtml
Membership application: genetics.faseb.org/ashg07/index.html
Dues: $150

ACMG: www.acmg.net/members/application.asp
Membership application: www.acmg.net/members/application.pdf
Dues: $170

1. Epstein CJ. Medical genetics in the genomic medicine of the 21st Century. *Am. J. Hum. Genet.* 79:434-438. 2006.

2. Elaine Strass, Executive Director, ASHG. Personal communication (December 2006).

3. Genetic counselors comprise about nine percent of the active ACMG members, but this group represents less than seven percent of the full NSGC members. ACMG, personal communication (January 2007).

Plea for ASHG and ACMG Involvement for NSGC Members

Dear Editor:

I am writing in support of **Judith Benkendorf's** letter, "A Sad Day for the Genetic Counseling Profession." I share Benkendorf's concern regarding the diminishing membership of genetic counselors in ASHG and our diminishing presence at the national genetics meetings.

I believe that two events hastened this trend. The first was the decision in 1993 to discontinue having NSGC and ASHG meet back-to-back in the same location. I was in the minority in voting for the continuation of our meeting together because I feared the time when fewer genetic counselors would attend ASHG. The second event was the establishment of the American Board of Medical Genetics (ABMG) and the resulting schism between the MD geneticists and genetic counselors. At the time, I was vocally in opposition of the ABMG, but I came to understand that it was the only avenue to ensure that the MD training programs in genetics would be counted among the medical specialties.

In 1991, the year the American College of Medical Genetics (ACMG) was established, I was asked to become a member of the Education Committee. I agreed because it offered an opportunity to work with genetic MDs and PhDs. I am currently serving on both the Membership and Program Committees of the ACMG and enjoy the interaction among the genetics professionals.

As Benkendorf states, genetic counselors currently make up only four percent of the ASHG membership and represent only nine percent of the ACMG membership. Genetic counselors provide the majority of clinical care in genetics and must have visibility at ASHG and ACMG to showcase our expertise. We are the genetic professionals who translate and synthesize the amazing scientific advancements in the field to the public.

I have listened to genetic counselors express the Rodney Dangerfield refrain of "getting no respect." We need to be at the table to garner respect. We need the visibility in the broader genetics community. We need to have our voices heard and counted. Too often I hear my colleagues ask, "What do I get for my membership fees?" We all know, in life we get what we give.

I urge the genetic counseling program directors to forcefully encourage students to join NSGC, ASHG and ACMG as a professional responsibility. Supervisors at all levels also should promote membership. Genetic counselors interested in committee membership in ASHG and ACMG should contact the genetic counselors on the various committees. Our roles expand, modify and adapt. I am constantly in awe of the talents I see in genetic counselors. Let others see our expertise and commitment.

Michelle Fox, MS
Adjunct Assistant Professor, Pediatrics
David Geffen School of Medicine at UCLA
Los Angeles, CA