PERSPECTIVES
in genetic counseling

Volume 26 Number 1 Spring 2004

national society
of genetic
counselors, inc.

nsgc

the leading voice, authority and advocate for
the genetic counseling profession

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NSGC acknowledges Women's Health Care Services of Wichita KS, for a sponsor's educational grant to support this newsletter.

Women's Health Care Services, providers of late abortion care for fetal anomalies, George R. Tiller, MD, Medical Director. 800-882-0488.

'04 - '06 Strategic Plan Nears Completion

Bea Leopold, MA

As we go to press, the final draft of our 2004 - 2006 Strategic Plan is being voted on by the Board of Directors. (See related articles, pp 2 & 3.)

If approved, this plan will focus on three strategic initiatives:

1. Genetic Counseling Service Delivery Model
2. Billing and Reimbursement
3. Visibility.

Each initiative includes:

- Issues that define the topic
- Goals that answer the question, "What will we do?"
- Tactics that answer the question, "How will we do it?"
- Outcomes that give us a measure and timeline
- Lead Teams that assign responsibility.

Membership Involvement

How can you help? Several items call for membership discussion. In the next several months, we will initiate a dialogue with all members about the issues of nondirectiveness and scope of practice. These dialogues will take place over the listserv, in *Perspectives,* at Regional meetings and on CommuniShare. We are hoping to dedicate an issue of our *Journal* to nondirectiveness next year. Finally, all members are invited to contact anyone on our Board with comments.

We will post our strategic plan on our website once it is approved. We invite all of you to read it and join us in giving focus to these three vital issues. ❖

☞ www.nsgc.org

Palliative Care for Genetic Disease

A "Comfort" for Patients and Families

Lori S. Farmer, RN, ARNP, MS, APNG

In the last 50 years, genetic technology has provided clinicians with increasing diagnostic capability, resulting in comprehensive tools and treatments to improve the quality of life or extend the life span associated with genetic disorders. At the same time, little attention has been paid to the role of genetics professionals working with patients who have no treatment options or who are facing the end of life.

Palliative Care Defined

In 2003, the World Health Organization defined Palliative Care as "the active total care of patients whose disease is not responsive to treatment." This differs from End of Life Care, which refers to care in life's final stages. Genetic professionals should understand the difference between these terms, as palliative care aims not only to relieve the symptoms of incurable disease but to help patients and families create a meaningful experience from the point of diagnosis through the end of life.

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PRESIDENT'S BEAT

When most of us think of strategic plans, we envision well-written objectives with measurable outcomes that help to guide an organization into the future. The actual strategic planning process, however, is not as smooth as you may think from reading the final plan. Your Board of Directors spent the last weekend in January discussing and dissecting key issues that need to be addressed in our 4th strategic plan, one that will span through 2006. It was a weekend filled with peaks and valleys, frustration and enthusiasm, regression and progress. I saw amazing teamwork, ideas, dedication and, above all else, passion. I am thrilled to say that we have more than a written document.

> ***"Through your participation in the environmental scan and committee work, your voices were heard."***

Perspectives in Genetic Counseling is published quarterly by the National Society of Genetic Counselors, Inc. Send articles and correspondence to the Executive Office.

- EDITOR: Jessica Mandell • jmandell@mail.slc.edu
- DESKTOP PUBLISHER: Anne Greb • agreb@genetics.wayne.edu
- STAFF: Susie Ball; Janice Berliner; Jennifer Claus; Shelly Cummings; Kathleen Fergus; Nicole Fernandez; Angela Geist; Katherine Hunt; Sarina Kopinsky; Melanie McDermet; Kathy Morris; Whitney Neufeld-Kaiser; Claire Noll; Karen Potter; Roxanne Ruzicka; Faye Shapiro; Kathryn Spitzer Kim; Kathryn Steinhaus French; Beverly Yashar
- NSGC EXECUTIVE OFFICE: c/o Bea Leopold, Executive Director, 233 Canterbury Drive, Wallingford PA 19086-6617; &610-872-7608; FYI@nsgc.org

The opinions expressed herein are those of the authors and do not necessarily reflect those of the editorial staff or NSGC.

*Next issue **June 15***
*Submission deadline **May 11***

We have a living, breathing strategic plan that will move NSGC closer to realizing its mission of obtaining recognition of the genetic counseling profession as an integral component of health care delivery, education, research and public policy. I want to take this opportunity to give you a preview of the issues that your BOD identified as strategic plan priorities.

GENETIC SERVICE DELIVERY

Taking a global look at genetic service delivery models is a primary initiative of our strategic plan. Based upon data from our environmental scan, it is clear that NSGC needs to initiate efforts to define the genetic counselor's scope of practice, assess existing and potential service delivery models and replace/redefine nondirectiveness as the central tenet of genetic counseling. I am well aware that many counselors may object to or be leery of the potential impact these endeavors have on our clinical practice. However, if NSGC does not initiate this effort, these issues will be decided for us by others, which may not necessarily be in the best interests of our profession. We must address these topics ourselves. Genetic counselors must recognize that aspects of our practice can, will and should be done by other health care providers. Our challenge is to identify what we do that is unique, define the value of our service in health care delivery and market that value to other health care providers.

VISIBILITY

Visibility is another strategic initiative that we defined. As I stated in my presidential address, increased visibility of NSGC is vital if we are to continue to be the leading voice, authority and advocate for our profession. Our environmental scan data reveal that our organization needs to be less insular and reach out to non-genetic health care professionals. NSGC must launch a targeted public relations campaign to identify and form strategic alliances with other health care providers. In addition, NSGC actively must identify and join issue-based advocacy coalitions in order to stay abreast of and influence legislative policy.

BILLING AND REIMBURSEMENT

Billing and Reimbursement is once again a key strategic initiative. It is not our intent to solve our B&R problems by 2006, but NSGC will launch efforts to reach our goals. We will act on the recommendations made by our B&R Task Force. NSGC will conduct an outreach campaign to national third party payors, develop a tool kit with strategies to implement B&R on local levels and continue to gather data and support research regarding B&R.

YOUR VOICE COUNTS

This outline gives you a glimpse of the overall initiatives of our strategic plan. The final strategic plan document will soon be available to all members. I urge everyone to remember that although your Board hammered out the details, the plan was written by all of you. Through your participation in the environmental scan and committee work, your voices were heard. In addition, if NSGC is to realize the strategic plan's goals and tactics, it will take all of our efforts on local and national levels. Please contact your region representative, a committee chair or an executive committee officer to get involved in these efforts. Together we can move NSGC toward a bright, successful future. ❖

Dawn C. Allain

Dawn Allain
2003-2004 President

BOARD OF DIRECTORS HIGHLIGHTS: DEVISING OUR STRATEGIC PLAN

Cathy Wicklund, MS,
NSGC Secretary

In January, NSGC's Board of Directors convened to focus on our 2004 – 2006 strategic plan. This intense meeting contained two full days evaluating our 2001 – 2003 strategic plan, exploring trends among other associations and prioritizing strategic initiatives for the next three years.

ASSESSING OUR HISTORY

Bea Leopold presented a history of the development of our past strategic plans. The Board then evaluated our 2001 – 2003 plan, including ratings on a three-point system of how well we addressed each of our major goals. Our high-level initiatives were Web Enhancement, Public Relations and Expansion of our Executive Office; our mid-level initiatives were Billing and Reimbursement and Legislative Issues; our lowest-level initiatives were Expanded Roles and Global Awareness.

ACKNOWLEDGING SUCCESSES

The Board acknowledged the following NSGC successes in the past three years:

- Our PR firm developed a video news release to generate media stories and interest about genetic counseling.
- Our input was solicited in private, corporate and governmental venues.
- Website stats show that we are receiving 20 – 30,000 homepage "visits" per month. This number translates higher when compared to "hits," which identifies direct-link entry anywhere on a website.
- We reached a $1 million budget and have hired a firm to conduct a Billing and Reimbursement study.

TRENDS

Each Board member read extensive material published by the American Society of Association Executives' Foundation about external and internal trends affecting associations today. The Board identified the following external trends (factors beyond the scope or control of NSGC or its members) that may impact NSGC in the next several years:

High Impact Trends:
- Economics
- Science and technology
- Social changes

Middle Impact Trends:
- Demographics
- Work/Workplace

Low Impact Trends:
- Global
- Business
- Government

Impact of these trends may include how we define genetic counseling practice, our delivery of genetic services, the shift of genetics in physician offices and the need to educate physicians, insurers and human resource professionals on the value of our services.

The Board then discussed internal (member-driven) trends that may affect our organization and our future. Most notable were the aging-down of our membership, the skew of 46% of our membership working in the field less than five years and the diversification of work settings. We reviewed the positive and negative effects of licensure and the segmentation of our members as we grow into new fields. We also looked at member preferences, as expressed in the Internal Environmental Scan. Our top member benefits, in usefulness order, are: Annual Education Conferences, Membership Directory, Listservs, *PGC*, Professional Status Survey and *JGC*.

CONSULTANTS LEAD CRITICAL DISCUSSION

Consultants from The STAR Group facilitated a discussion about the environmental scan and setting priorities for the next strategic plan. We were cautioned to focus clearly on what issues can be realistically achieved in a three-year timeframe. Our top priorities for the next strategic plan are: defining our scope of practice, assessing and exploring other genetic service delivery models, addressing Billing and Reimbursement and increasing visibility. Bea and **President Dawn Allain** were charged with drafting a strategic plan, based on these goals. A vote on the final document is pending at presstime.

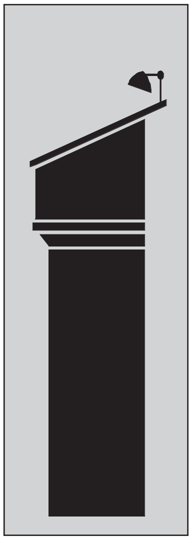
Our top member benefits, in usefulness order, are: Annual Education Conferences, Membership Directory, Listservs, PGC, Professional Status Survey and JGC.

OTHER BOARD BUSINESS

Consultants **Barbara L. Ciconte** and **Eugene A. Scanlan, PhD,** of Donor Strategies, Inc., an independent research firm located in Chevy Chase MD, presented a brief overview of their study on whether a foundation is financially feasible for NSGC at this time. Information from two breakout groups at our Board meeting will be added to their research, which includes interviews with members as well as corporate and foundation community leaders.

Kelly Ormond, President-elect, updated the Board about the Governance Task Force. This team will explore the current structure and function of the Board of Directors to best meet the needs of the Society.

The board will reconvene in early June in Chicago. ❖



Palliative Care for Genetic Disease, *from page 1*

How then should conditions such as cystic fibrosis be approached when life expectancy has increased from 15 years in 1977 to 33.4 years today, or when patients with inherited, adult onset diseases are living longer due to better screening and medications? Regardless of the diagnosis, most individuals with an incurable disease desire to minimize pain and distress and want family members, friends and health care providers to support their psychological, social, spiritual, physical and economic needs. When such care falls short, patients often face a technologically over-treated and prolonged death. Genetics professionals, and all health care providers, should be obligated to provide the best possible care for seriously ill patients, including advocating for the relief of suffering and being part of a team to help facilitate a death that is congruent with the values and desires of the patient and family.

Help From ISONG

A 2002 survey of participants at the International Society of Nurses in Genetics (ISONG) helped to assess palliative care issues (Farmer, L. and Lipe, H.). Of respondents, 42.8% felt that patients with a life threatening genetic diagnosis should receive palliative care as soon as the condition is diagnosed, while 23.2% felt care should be initiated after all reasonable curative efforts had been exhausted, and 3.5% felt that palliative care is appropriate only in the last few months of life. Potential problems that could impede the provision of palliative care included: lack of education by health care providers, inadequate financial resources at a medical facility and concerns about patients' financial resources to pay for palliative care.

These results identify obstacles but also some directions. To enhance palliative care in genetics, NSGC, ISONG and other genetics groups could review the "Last Acts Precepts of Palliative Care," a document developed by the Task Force on Palliative Care in 1997, now adopted by over 300 health care organizations. This document examines symptom management among patients, patient/family decision-making and assistance with communication, coping and loss.

"COMFORT" Framework

Individual genetic counselors also can help organize a health care team at their facilities dedicated to the Palliative Care Interventional Framework, designated by the acronym COMFORT^:

Call in the entire health care team to develop a palliative care plan for the patient and family.

Open lines of communication between the patient, family and medical staff.

Make the team available as the patient's condition changes over time.

Facilitate the patient's and family's right to exert control before, during and after death.

Optimize resources to assist the patient and family.

Revise the care plan as needed.

Think about the "end" in the beginning. Death is a process, not an event!

Genetics nurses, counselors and other health professionals have a responsibility to appreciate patients diagnosed with incurable or life threatening genetic disorders and to advocate for comprehensive palliative care. Allowing every patient an excellent quality of life is a goal that will enrich, inform, enlighten, support and sustain us, as patients, families and caregivers. ❖

Professional Status Survey Success!

***Kristen Mahoney Shannon, MS** and*
Cecilia Fairley, MS

It's fair to say that most genetic counselors anticipate the publication of the bi-annual Professional Status Survey (PSS). NSGC members anecdotally report that they have used the data to increase salaries and negotiate for conference reimbursement. Read one such story, and learn how a group of genetic counselors at Children's Memorial Hospital in Chicago really got something to celebrate!

Shortly after the 2002 PSS data was presented at the Annual Education Conference, **Cecilia Fairley** asked that the data be queried for salary ranges and medians of genetic counselors practicing in the Chicago area. The data on the PSS isn't collected by city, but it could be analyzed for the whole state of Illinois. Cecilia and her colleagues presented the Illinois salary analysis to their human resources department in March 2003. This prompted a salary review with other hospitals in the region (since the counselors at Children's were at the very bottom of the pay scale, according to the analysis). The hospital agreed that the counselors needed a pay increase to meet the minimum of other local institutions.

Since that time, counselors have received up to a 10% raise dependent upon their years as employees. The counselors intend to submit the next PSS results to their human resources department again to assess their standings with that of their colleagues in the state of Illinois.

This story illustrates the importance and benefit of the PSS. The next PSS will be administered in the summer of 2004. Make sure you participate! ❖

EVALUATING GENETIC RESEARCH STUDIES

Kelly A. Taylor, MS, Co-chair, Research Subcommittee

Genetic research has rapidly moved from a focus on single-gene disorders to the study of complex disorders like multiple sclerosis, diabetes and cardiovascular disease. As clinical genetic testing for these disorders is often unavailable, participation in research is growing. Genetic counselors must assess the quality of genetic research to help clients make informed decisions about whether to participate. Here are some points to consider when evaluating a research study.

CONFIDENTIALITY

How will the confidentiality of samples and information be maintained? Access to identifiable information should be secure and limited to individuals with a "need to know." Paper files should be kept in locked cabinets or rooms, and electronic information should be stored in password-protected databases. Samples should be coded without names, date of birth, medical record number or research data, if possible. Only anonymous or coded information should be shared between collaborators, and research should remain separate from clinical charts. The genetic counselor should determine if a Certificate of Confidentiality has been issued by the National Institutes of Health which allows researchers to protect identifying information from third parties even if subpoenaed.

USE OF SAMPLES

How will samples and information be used? Some studies send samples to a repository outside of the investigator's institution, available to public researchers. Many investigators also want to use samples for future studies. Participants should choose whether or not their samples can be sent to a repository or used for future, unspecified studies. If participants decide that they no longer want to participate in a study they should be given the option of having their samples, and any personal data, destroyed or anonymized.

RESULT DISCLOSURE

Will individual results be provided to participants? If results are offered, the genetic counselor must assess whether their significance has been confirmed and if the utility of the test has been established in predicting risk, confirming the diagnosis or making medical management decisions. The genetic counselor should determine if the lab is CLIA approved and whether or not genetic counseling will be provided by the study. If results are not disclosed, the genetic counselor should find out what would be done if medically compelling results were found for a participant.

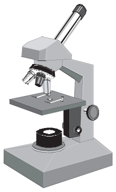
RESEARCH UPDATES

Will general results be provided? Some researchers offer updates on the findings of their studies through newsletters or websites. Even if participants never learn individual results, being informed of the overall findings of the study may affect their decision to participate.

According to the Genetic Alliance in its Consumer Indicators of Quality Genetic Services, "Information about genetic research is available to the consumer and integrated in clinical practices." As such, providing information about genetic research is a key aspect of quality health care, and genetic counselors with knowledge of research studies fulfill a necessary service for

RESEARCH NETWORK

U.S. FOUNDER DELETION IN MSH2 AMONG HNPCC FAMILIES



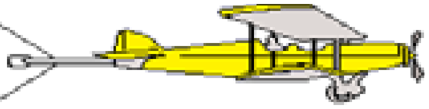
Ohio State University and Creighton University recently reported nine seemingly unrelated families that share the same exon 1 – 6 deletion of the MSH2 gene [*JAMA*. 2004 Feb 11;291(6):718-24]. Further haplotyping proved the families are related, and genealogy work has connected three families directly to a couple that immigrated to the United States from Germany in 1727. These families first settled near Philadelphia, and then some moved to Guilford County in North Carolina. From there, members began moving westward through the Cumberland Gap as far as California.

To determine the proportion of HNPCC mutations in the U.S. that are due to this single mutation, we are seeking the following:

1. **Families with a known exon 1 – 6 deletion of MSH2**
   Molecular analysis will be performed to confirm that the breakpoints are the same as those founder families. Free, CLIA approved testing will be offered for at-risk relatives, provided the family releases their results and medical and family history and participates in the geneaology work.
2. **New families with the American founder deletion of MSH2**
   Molecular analysis will be performed on patients with MSI+ tumors who have absent MSH2 staining on IHC and have sequenced negative for MSH2. Anyone meeting the Bethesda criteria who is in the migration path of founder families may also be considered for testing. ❖

☞ Heather Hampel, hampel-2@medctr.osu.edu, 888-329-1654

Media Watch



Angela Geist, MS and Roxanne Ruzicka, MS

November 19 – 940 KGMS, *Straight Talk*

This AM talk radio program interviewed **Donita Vogt** about the Pregnancy Risk Line in Arizona which serves as the national headquarters for the Organization of Teratology Information Services (OTIS). The show gave a great overview of our profession.

December '03 – *American Baby*, "Should you see a genetic counselor?"

This article discusses the indications for, and the process of, genetic counseling. **Vivian Weinblatt**, **Rene Chard** and **Allison Gregory** are quoted. NSGC and its website are noted. The article is generally accurate, but there are a few errors, such as, "If you're already pregnant, you and your spouse will give blood samples, which are then tested for the diseases you may potentially carry."

December 3 – *Boston Globe*, "Working the Odds"

This article features **Kristen Niendorf** and the job of cancer genetic counseling. The writer describes the psychological issues and uncertainties involved in genetic counseling.

December 3 – *Wall Street Journal*

The story focuses on the increase in predictive testing for disorders like Alzheimer disease, breast cancer, colon cancer and diabetes. **Susan LaRusse** notes that despite limitations in these tests, many people with Alzheimer disease in their families request the apoE gene test. The article also states, "To help patients wrestle with the emotional and medical implications of the information genetic tests provide, medical centers have rushed to add genetic counselors to their staffs. There are 2,200 genetic counselors working in health centers, up from 1,000 ten years ago. They help patients determine whether they should get a test, interpret the results and answer questions about treatment, even insurance coverage." A link for genetests.org is provided.

December 4 – CBSNews.com, 60 Minutes II, "The Heart Attack Gene"

A large family from Iowa with a strong history of heart disease is portrayed as possibly holding the key to stopping heart disease in its tracks. Linkage studies at the Cleveland Clinic identified a genetic mutation thought responsible for the heart attacks. Results were also published in *Science*. The story discusses how some family members are concerned about genetic discrimination. Relatives who carry the mutation were advised to seek preventive medicine and watch their diet and exercise.

January 6 – PBS, Scientific American Frontiers, "Friendly Genes"

A story about Williams syndrome (WS) is hosted by **Alan Alda**. He attends a WS support group picnic and interviews both affected children and adults. The PBS website reports on statistics and symptoms of WS.

January 10 – Medscape.com, "Genomics and Managed Care: Preparing for the Revolution"

This informative article defines the genome and how increasing knowledge will effect health care and insurance. The genetic counselor's role in explaining and coordinating genetic services is described.

January 23 – ABCNEWS.com, "Making Better Babies"

This report covers a new study in *Lancet* suggesting that all women, regardless of age, could benefit from prenatal tests such as amniocentesis. The study examined the costs and benefits of testing and found no relationship between the benefit of testing and the mother's age or risk status. The article correctly states that doctors generally recommend genetic testing if there is a positive family history or an abnormal ultrasound or if the woman is over 35 years of age. Researchers raise concerns about opportunities for abuse, such as selective abortion based on gender.

January 24 – *The Houston Chronicle*

A very long article follows a 13-yo with cystic fibrosis (CF) who received Houston's first lung/liver transplant and seems to be doing well. The article does not mention genetic counseling but has a blurb about how CF is inherited.

January 26 – *Wall Street Journal*, "Alternatives to Amnio"

This article reviews prenatal screening methods and diagnotic procedures such as CVS and amniocentesis. Most of the information is factual, though it describes amniocentesis as a "30 to 50 minute procedure." It is not clear that the reporter understands the difference between detection rate and reliability of results. Genetic counseling is not mentioned.

February 9 – *Reno Gazette-Journal*, "Preemptive Strike"

A woman chooses to undergo a prophylactic mastectomy and hysterectomy after watching several family members die of cancer and after testing positive for BRCA2. **Robbin Palmer** is quoted.

February 16 & 23 – FOX-TV, "The Littlest Groom"

A two-part reality show based on "The Bachelor" featured a 4'5" young man searching for a bride who had to choose between little women and women of average stature. ❖

Results…Video News Release

STAR/Rosen reports that in the six week period between January 26 and March 8, our Video News Release was aired on 40 stations with an audience reach of nearly 900,000 viewers. It was aired on all major networks. ❖

2004 Conference Update

WELCOME (BACK) TO WASHINGTON DC!!

Barbara Willis Harrison, MS, Logistics Chair

In 1790, the site of our nation's capitol was chosen by Congress as a natural midpoint between Northern and Southern states. An area along the Potomac River "ten miles square" was laid out by African-American mathematician **Benjamin Banneker** and surveyor **Andrew Ellicott**. French engineer **Pierre Charles L'Enfant** was hired to design the city, which was to be the model of a democratic capitol.

Fast forward to 2004. Washington DC has it all – historic monuments, breathtaking museums, great restaurants, exciting nightlife, amazing diversity and beautiful parks! When you come to Washington for our next AEC, we want to make sure your stay is productive, stimulating and fun, and we have many local activities in store, in addition to the learning opportunities.

SHOPPING, SHOPPING!

As a starting point, The Hyatt Regency on Capitol Hill is located next to magnificent Union Station, the most visited site in all of DC. Union Station is the most exciting and dynamic shopping destination in the country, and the quality and diversity of its stores have made it a favorite of Presidents as well as millions of commuters and tourists.

TOURING THE TOWN

Several exciting tours leave from Union Station on a regular basis. These include the Old Town Trolley Tour, featuring such attractions as the National Cathedral, Lincoln Memorial, Georgetown, the White House and the museums of the Smithsonian. The tour has "on and off" privileges, so you can personalize your day to allow extra time at the places you desire. There is also DC Ducks, a popular tour over land and water. Lastly, a unique tour, Monuments by Moonlight runs at night and covers over 100 points of interest. Each of these tours currently cost $28.

RELAX AND REFRESH

A novel effort we are planning this year is to include some devoted "me" time. The AEC provides us many opportunities to increase our knowledge, converse with colleagues and enhance areas of our practice. However, it is common to feel overwhelmed and exhausted during the conference. To counteract this, we are planning "wellness" activities to help us regain our focus. During lunch breaks and in the early mornings and late evenings, we will offer massages, yoga and meditation and walking/running groups. The 2004 AEC Planning Committee wants to stimulate your mind, but we also want you to take care of yourself!

Look for more information on how to sign up for tours and wellness activities in the AEC brochure, which will be mailed in March. ❖

Abstract Deadline is June 4th!

'04 Short Courses

Two Short Courses will be featured prior to our Annual Education Conference in Washington DC. Both are scheduled for October 6 and 7.

EXPANDING OPPORTUNITIES IN RESEARCH

The complex and unique medical, ethical and social issues associated with genetic research studies have created a wide variety of novel job opportunities for genetic counselors. This course will address the essential elements and skills necessary for these roles: monitoring and ensuring compliance with genetic research regulations, writing research protocols and consent forms, interacting with Institutional Review Boards (IRB), appropriately identifying, recruiting and interacting with research participants and researching and incorporating ethical and legal components to genetic research.

THE ABC and D'S OF NEUROGENETICS: HD, AD, PD AND BEYOND

The purpose of this course is to provide an educational and interactive forum for genetic counselors to learn about complex neurogenetic diseases with specific focus on the practice of genetic counseling. Our goals include: to improve knowledge of Mendelian and complex trait neurogenetic diseases including etiology, clinical features, diagnosis and treatment; to provide participants with a primer on neuroanatomy, neurological exams and frequently used neuroradiological and neuropsychological tests and technology; to present the psychosocial difficulties encountered by individuals and families coping with these conditions; and to discuss current and future ethical issues in neurological genetic counseling. ❖

RESOURCES

CHOOSING NAIA

***by:* Mitchell Zuckoff**
Beacon Press, Boston MA, 2002,
301 pp., $16.00 (paperback)
Reviewed by: Elyse M. Weber, MS

Choosing Naia chronicles the story of **Greg** and **Tierney Fairchild** who learn during their pregnancy that their daughter, **Naia**, has Down syndrome. This story originated as a series for the *Boston Globe* about prenatal choices and was picked up by the popular media including NPR and ABC News. Struck by the strength and determination of this family, **Mitchell Zuckoff** decided to expand the story into a book.

A TRUE STORY

Zuckoff tells much of the story through quotes from the Fairchilds, an educated and financially stable couple. When a second trimester ultrasound detects a congenital heart defect, amniocentesis confirms their fetus has Down syndrome. As the couple decides whether to continue the pregnancy, they speak with family, medical specialists and parents of affected children. They elect to continue the pregnancy, and Naia is born by emergency C-section.

Over the years, the Fairchilds shift their focus from the early months of doctors visits to the discrimination Naia faces. Naia's mental development is discussed, and the couple navigates the school system. Tierney becomes pregnant again and does not have an amniocentesis. The couple's second child, a son, is born without Down syndrome.

The Fairchilds possesses a great deal of personal insight, due in part from their experiences as an interracial couple (Tierney is Caucasian, Greg is African American). The couple's own experience of discrimination helps them advocate for their daughter and others with disabilities.

THE BIGGER PICTURE

Zuckoff interviews genetic counselors, obstetricians, cardiologists, nurses, physical therapists and families with Down syndrome children. Stories from couples who terminated an affected pregnancy create a book surprisingly neutral on the issue of termination.

Genetic counseling is favorably portrayed. **Alicia Craffey,** who connected the Fairchilds and Zuckoff and was interviewed for this project, plays a prominent role in the couple's decision to continue the pregnancy.

SIDE-NOTES

Additional side-notes cover the eugenics movement and sterilization, historical theories of the causes of mental retardation, institutionalization and plastic surgery to "fix" the facial features in Down syndrome.

Zuckoff's descriptions of the maternal serum screen, prenatal diagnosis and non-disjunction are accurate, and the history of genetics and social issues are interesting. References are given.

For genetic counseling students, this account is helpful in grief processes, decision-making and giving bad news and the experiences families have outside of our offices. Nurses, obstetricians and pediatricians also could learn from this book. ❖

A TALE OF TWO COUNSELORS

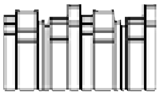
Ethics Subcommittee

GENETIC COUNSELOR JANE has an ethical question about one of her cases and needs a response in a hurry. She tries the listserv but still isn't satisfied. She remembers that the Ethics Subcommittee offers informal and formal consults. For both services, the genetic counselor submits a case by email or phone. The Subcommittee discusses the case, and a member reports back to the requesting genetic counselor. Turnaround for an informal consultation takes about a week. A formal consult is more detailed and takes more time. In both consults, the dilemma is addressed and the Code of Ethics is applied. Genetic Counselor Jane decides to submit her case for an informal consult to the Subcommittee.

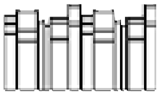
GENETIC COUNSELOR JOE is having some conflict with one of his co-workers that is impinging on patient care. He contacts the Ethics Subcommittee for a formal consult. The Subcommittee reviews the case and notes that in the Code of Ethics, genetic counselors should encourage ethical behavior in their colleagues. The Subcommittee addresses the behavior of the colleague in question and the relationship between the colleague and Genetic Counselor Joe. A formal written summary is sent to Genetic Counselor Joe citing the ethical dilemmas posed and noting the sections that apply. With the Ethics Subcommittee's input, Genetic Counselor Joe is now empowered to improve his work situation.

If you have a case that you think may benefit from an ethics consultation, please contact a Subcommittee member. That is what we are here for! ❖

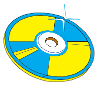
Kim Hart, Chair	khart@bmrn.com
Melinda Cohen	melinda.cohen@mcmail.vanderbilt.edu
Jill Goldman	jgoldman@memory.ucsf.edu
Erynn Gordon	egordon@cnmcreasearch.org
Michelle M. Moore	michelle.moore@memhospcs.org
Roxanne Ruzicka	RoxanneR1@aol.com
Kelly Taylor	ktaylor@phg.mc.vanderbilt.edu



RESOURCES



SOMEHOW WE'LL MAKE IT WORK



by: **Ellen C. Perrin, MD**
and **Jennifer Schott, MD**
Video and/or CD, $75

WHAT DO KIDS THINK? THE DEVELOPMENT OF CHILDREN'S CONCEPT OF ILLNESS

by: **Ellen C. Perrin, MD**
Video only, $50

Reviewed by June A. Peters, MS

Two new videos, produced by doctors in Boston, offer insight and teaching tools regarding patients and families facing children's illness.

PERSONAL ACCOUNTS

Somehow We'll Make It Work is an hour-long presentation of interviews with chronically ill children and their relatives. The video is broken into sections by themes, such as diagnosis, adaptation and family re-organization. We witness families transition from shock to grief to adjustment as the illness becomes part of their lives.

While the diagnoses in the video are not all genetic, many of the issues remain universal. The families recognize a conundrum — they do things that may not be normal for other families but help them live as normally as possible. For example, some kids with cystic fibrosis keep digestive enzyme pills at friends' houses in case they want a snack so that they won't feel left out. The video also explores family relationships. Parents share how they drew closer or became more distant as they tried to cope. Many families find inspiring ways to be optimistic and clarify their priorities.

A PROFESSIONAL TOOL

How Do Kids Think? is a tool for health care professionals about framing interviews and education for well and ill children. Psychologists and pediatricians conduct interviews with children to illustrate asking open and yet specific questions such as, "How do you know when a person is sick?," followed by probes to draw out further information.

Such tools help to assess a patient's functional level of understanding. Children with serious illnesses may appear sophisticated because they use correct medical language. However, their true understanding of illness may be low. A genetic counselor, or other provider, should assess all children's understanding of information and take time to explore concepts fully.

RECOMMENDED FOR EDUCATION

I would recommend these tapes for use in training genetic counselors, health professionals and students. They augment required readings about child development and family adaptation to chronic illness and can stimulate discussion and role-playing. ❖

☞ Ellen C. Perrin, MD, Center for Children with Special Needs, New England Medical Center, 750 Washington St., Boston MA 02111

NEWEST MEMBER BENEFIT A HIT!

Aubrey Turner, MS

According to statistics for the week of February 29, our newest website member benefit, the JobConnection Online, has risen to be among the top five visited pages on our site, making it a "hit" as a member resource! JobConnection Online went live on January 18.

In a broad sense, the top two reasons people visit our website are to find a genetic counselor and to explore a genetic counseling career/job. Although a flaw of these statistics is that visitors of the main JobConnection page are then likely to visit the job listing or job search pages, the numbers show that this new section of the website has been well utilized.

Rank	URL	Hits	% Total Hits
1.	/resourcelink.asp	4,127	18.29%
2.	/Index.asp	2,966	13.14%
3.	/members/index.asp	1,541	6.83%
4.	/TrainingProgram.asp	1,373	6.08%
5.	/resources/jconn/job_detail.asp	956	4.24%
6.	/careers/index.asp	859	3.81%
7.	/resources/jconn/listjobs.asp	786	3.48%
8.	/resources/jconn/searchjob.asp	516	2.29%
9.	/careers/howtobe.asp	338	1.50%
10.	/conference/index.asp	301	1.33%

Thanks to the Executive Office and ProWebConcepts for orchestrating this new website feature! ❖

INFO AVAILABLE: PRENATAL TRIPLE SCREEN AND X-LINKED ICHTHYOSIS

Low estriol levels on a prenatal triple screen in pregnant women can be a marker for X-linked ichthyosis. The Foundation for Ichthyosis & Related Skin Types can provide disease information, physician referrals, information on additional diagnostic testing and a support network for expectant mothers with a suspected X-linked ichthyosis fetus. ❖

☞ 800-545-3286,
www.scalyskin.org;
info@scalyskin.org

SIG ACTIVITIES

'04 CANCER SIG AWARDS

Karen Huelsman, MS, Chair, June Peters, MS** and **Monica Alvarado, MS

The Cancer SIG Grant Award Committee is pleased to announce the 2004 award recipients with funding of $2800. The Cancer SIG Grant Award was designed to maximize the use of annual SIG funds by giving members the opportunity to compete for surplus SIG money. CSGA recipients present the results of their studies at the NSGC Annual Education Conference or at the SIG meeting. Congrats to our recipents, and thanks to our grant review team for a super job!

$1100 – **Mollie K. Lyman, MS** and **Carolyn D. Farrell, MS, CNP**

DNA Banking: Saving for the Future, Roswell Park Cancer Institute, Buffalo NY

Diagnosis of a hereditary cancer syndrome may not be currently possible, yet future molecular genetic testing may offer help. Individuals and families frequently are not referred to cancer genetics until an affected relative is deceased or in the end stage of life. For these reasons, DNA banking is essential so additional genetic testing may be done in the future. This study will investigate the offering and uptake of DNA banking by genetic counselors and the understanding and utilization of DNA banking by other oncology practitioners. This information may enhance the recognition and utilization of DNA banking and help create guidelines.

$900 – **Katrina Lowstuter, MS** and **Randall Burt, MD**

Status of Hepatoblastoma Screening for At-risk FAP Patients, Huntsman Cancer Institute, University of Utah, Salt Lake City UT

Clinical cancer genetic team members will be surveyed on how screening for hepatoblastoma in at-risk FAP individuals is presently conducted. The survey will evaluate screening recommendations for FAP including frequency and appropriateness of screening. The study also will assess genetic testing at birth related to managing hepatoblastoma screening.

$800 – Jennifer Gamm, MS, Jessica Everett, MS and **Carrie Gill, BS**

Effects in Women from Direct-to-Consumer Advertising of BRACAnalysis®, Cincinnati Children's Hospital Medical Center, Cincinnati OH

Recently, Myriad Genetics piloted a direct-to-consumer campaign for BRACAnalysis®. While the targeted audience encompasses a minority of the population, the media campaign will reach many more women who may not have a predisposition risk for breast cancer. The impact of this commercial continues to be debated. This study will determine whether the commercial changes breast cancer anxiety in women and, if a woman's intent to pursue testing is affected, where she would go to seek information. ❖

☞ www.nsgc.org/members/sig/sig_familial_crc.asp

FOCUS ON INDUSTRY SIG

Lori Ann Correia, MS** and **Mary Freivogel, MS, Co-chairs

Genetic counselors are spreading their wings out of clinical and academic settings and landing in industry. The Industry SIG was created in 2001 with 35 members and has grown to 80. Members work in commercial laboratories, biotechnology and pharmaceutical companies, government, public health, private practice and clinical settings. The numbers of industry-related companies that employ genetic counselors are only surpassed by the varied responsibilities of the individuals working for these organizations.

Here are three interesting careers of industry genetic counselors:

- **Trisha Brown** is Associate Vice President and National Assistant Director of Genetics and Genomics at a commerical lab. She coordinates a national genetic counseling service, standardizes genetics operations and helps in business development, acquisition of new technology, marketing, CME coordination and development of Information Technology platforms for genetics.
- **Sylvia Au** works in government as a State Genetic Coordinator. She oversees the state's genetics program and is the principal investigator for several local and multi-state research projects. She develops public health programs such as newborn screening, coordinates funding for genetic services, implements legislation and genetic education, collaborates with the private sector and participates in state and federal policy development.
- **Kimberly Hart** is Clinical Research Associate (CRA) coordinating FDA clinical trials for mucopolysaccharide diseases at a pharmacogenomic company. She writes clinical protocols and informed consent forms, identifies new principal investigators with IRB submissions, audits study data and manages databases.

IMPACT ON THE FUTURE

The Industry SIG is privileged to have these genetic counselors, along with many others, in its membership. Their stories illustrate the broad professional opportunities in which genetic counselors can utilize their talents and skills. The Industry SIG hopes to increase interest in expanded counselor roles and in SIG activities. Watch the progress of our profession, and stay tuned for more articles about genetic counselors in industry. ❖

☞ www.nsgc.org/members/sig/sig__industry.asp

LETTERS TO THE EDITOR

RESPONSES TO ISSUES OF CERTIFICATION

ABGC BOARD: AN ESSENTIAL PROFESSIONAL STANDARD

The American Board of Genetic Counseling was founded in 1993 as an independent body dedicated to enhancing the professional development and recognition of genetic counselors. As current elected members of the Board, we would like to respond to the "Letters to the Editor" regarding certification in *PGC* 25(4):11, 2003.

Certification is a recognized mechanism by which professionals demonstrate minimum competency in their field. In health care, the goal of certification is to set

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A RESPONSE FROM THE FIELD

I am bothered by *PGC's* decision to allow a letter to the editor to be published anonymously ("Is Certification Really that Important?") and by the author's (mis)understanding of certification. Certification does not claim to assure excellence. It is a policy that verifies that the holder has met minimum requirements established by the profession. Policies sometimes exclude people who should be included, and vice versa. Genetic screening policies provide evidence for that. The anonymous writer states, "Many of us know board certified genetic counselors whose competence we question," but in the absence of certification, there would be no standardized measurement of competence at all.

If the ABGC allows "special consideration" as the author requests, it will weaken the certification process and diminish the accomplishments of those who hold certificates. I was on the committee that developed the ABGC over 10 years ago and on the original Board of Directors. I would oppose strongly any attempt to fix a process that isn't broken. A counselor who is not certified by ABGC is not necessarily a poor counselor; he or she simply is uncertified. It is reasonable from an employer's point of view to look first among job applicants who posses evidence of higher qualification, i.e., certification, 3-5 years experience, someone who is multilingual, etc.

Specific misconceptions that the writer presented include:

- NSGC excludes uncertified counselors from leadership positions. Our bylaws make no distinction among its members with regard to board certification.
- Uncertified counselors are excluded from some NSGC studies. A properly conducted study controls for variables, of which board certification may be one. Excluding non-certified counselors from a study is at the discretion of the study organizers.
- NSGC excludes uncertified counselors from some awards. The JEMF award is unavailable to uncertified counselors. However, as a founding member of the JEMF Board, I can report that this award requirement was set by the award's benefactor. Uncertified counselors are welcome to apply as co-investigators.

Unless a certified counselor chooses to add "CGC" to their title, I am not generally aware of which counselors are certified or not. It makes no difference to me. Perhaps much of the "stigma" associated with being uncertified is self-imposed. ❖

— Edward M. Kloza, MS

NSGC COMMENTARY ON CERTIFICATION

I would like to respond to the letter, "Is Certification Really that Important," *PGC* 25(4):11, 2003. NSGC is an organization that represents *all* masters-level genetic counselors regardless of their board certification status, and member benefits are based only upon membership status (full, associate, student, emeritus).

ALL MEMBERS ELIGIBLE FOR BENEFITS

With only one exception (the Jane Engelberg Memorial Fellowship), ABGC or ABMG board certification is not required to receive any NSGC sponsored award. This holds true for NSGC leadership positions as well. Certification is not required to serve on the Board of Directors, to be a SIG Chair or to be on a committee or taskforce. Lastly, NSGC has never funded a research study that required a participating member to be certified.

CONSISTENCY IN LICENSURE

Regarding board certification in licensure, most states require an exam as an objective measure of competence, regardless of the profession under evaluation. NSGC supports the utilization of the ABGC board certification process in licensure because it provides a consistent assessment of genetic knowledge and counseling skills. We anticipate that this will enhance reciprocity between states. The use of this examination also may keep licensure costs down, as states will not have to develop individual exams. We encourage states to work closely with all of their NSGC members to proactively address licensure issues.

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national society
of genetic
counselors, inc.

nsgc
*the leading voice, authority and advocate
for the genetic counseling profession*

233 Canterbury Dr Wallingford PA 19086

BE IN THE KNOW!

Information for applications and submissions associated with all of these deadlines are online and linked from our homepage: www.nsgc.org

April 5 - June 4 — Call for Abstracts, 23rd Annual Education Conference

April 30 — Call for 2004 – 2005 Board Nominations

April 30 — Call for Leadership Award Nominations

May 3 — JEMF grant proposals

May 15 — AHSPA grant proposals

☞ **IMPORTANT NOTICE: Future issues of *Perspectives* will be available online, only. Check out the system with this issue, too!**

LETTERS TO THE EDITOR

ABGC RESPONSE TO LETTER, *from page 11*

standards that lead to quality patient care. Certification and/or licensure is required of virtually all health care providers. This process requires satisfactory performance on specific examinations. It is not a perfect process; competent professionals sometimes experience difficulty with exams, and passing an exam does not guarantee that a professional is a good provider.

The ABGC certification exam is validated through the standard setting process used by the National Board of Medical Examiners. The process does not seek to fail a certain percent of individuals.. A specific score for every examinee is generated, and those who score above the determined cut off will pass.

ADDRESSING NEEDS

The ABGC is committed to uphold the standards of the profession and respond to the developing needs of genetic counselors. As such, ABGC:

- Has moved to two-year exam cycles to accommodate states seeking to use active candidate status for temporary licensure;
- Has developed a pathway for foreign-trained genetic counselors to obtain ABGC certification, modeled on the requirements for foreign-trained medical professionals practicing in the U.S.;
- Has established accreditation guidelines for genetic counseling programs. When employers hire genetic counselors with active candidate status or who are Board-certified, they can be assured the counselor has a minimum level of competence as determined by specific, objective measures;
- Is attuned to state licensure for genetic counselors. States will develop their own exams or use the ABGC certification exam. The ABGC will work with states to make necessary accommodations for non-certified genetic counselors to qualify to take the ABGC exam.

As a profession, we are establishing standards of practice for genetic counselors. Without these standards, others will establish them for us. Our goal is to be recognized by the public and the medical establishment as a legitimate profession with unique training and expertise. Patients have the right to be assured that their health care providers have met training requirements and demonstrated competence through an objective measure. To this end, we have a responsibility to ourselves and to the patients we serve. ❖

***T. Becker, MS, N. Callanan, MS,
L. Cohen, MS, A. Faucett, MS,
B. Finucane, MS, A. Greb, MS,
E. Reich, MS, D. Riconda, MS,
N. Warren, MS and L. Weik, MS***

NSGC RESPONSE, *from page 11*

On behalf of the NSGC Board of Directors, I assure the membership that we all contribute to the richly diverse group of genetic counseling professionals that make up our organization, and we strive to represent all members in policy decisions. Should you have concerns regarding these or other matters, I encourage you to discuss them with your representatives or with me personally. ❖

***— Dawn C. Allain, MS
NSGC President***