PERSPECTIVES
in genetic counseling

Volume 18, Number 1 Spring 1996

Cancer Susceptibility Counseling: Aspirations and Admonitions

by Beth N. Peshkin, MS, Georgetown Univ Med Ctr, Washington DC

Recent molecular studies have identified at least four major genes that contribute to breast, ovarian and colon cancer. The complex process of translating these discoveries into medical practice is proceeding rapidly. As technical issues are being resolved, we have an opportunity to reflect on the scientific and social implications of this new era of genetic medicine. This article defines areas where caution is needed and highlights aspirations for future practice and research.

Risk Assessment: An Emerging Science

■ **We must address key issues before testing becomes widespread.** Unresolved concerns include difficulties in interpreting tests, complex or restrictive protocols, long delays in obtaining results and cost prohibitions. Yet some commercial companies are already marketing genetic testing and computerized risk assessment directly to physicians. These pressures force us to examine the delineation between research and clinical applications.

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Positioning Ourselves for the Future: NSGC Formulates Marketing Plan

by Bea Leopold, MA, NSGC Executive Director

If a tree falls in the woods and no one is there to hear it, does it matter if it makes a sound? Long pondered by philosophers, this question applies to genetic counselors as well. If our work is not seen or appreciated, we too become lost in the woods. How can we position ourselves favorably, adjust to changes in the marketplace and keep an eye focused on future trends? Addressing these needs, the Board of Directors recently approved $15,000 to hire a marketing consultant.

Why Marketing?

"All genetic counselors, knowingly or not, market themselves every day," points out President Vickie Venne. Marketing is not simply advertising or selling yourself—it is making sure the image you project reflects the qualities you value.

NSGC's strategic plan recommends developing a marketing program. "This will allow us to define and create the tools necessary to accept the challenges of health care in the 21st century," asserts Venne.

NSGC's Priorities

President-Elect Ann Boldt is also a firm proponent of marketing. "It is imperative that genetic counseling be recognized as an integral part of health care delivery by the decision makers within managed care

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national society of genetic counselors, inc.

nsgc

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

On the Inside

NSGC acknowledges our following corporate friend for a grant to support this newsletter.

Presidential Remarks

Genetic Counselors Exemplify Our Worth

Puzzle pieces—my way of acknowledging individuals' contributions—are moving fast as NSGC members represent our field:

■ Karen Greendale's international activities have garnered an invitation to organize a session for the 1997 European Society of Human Genetics.

■ The Pedigree Standardization Project had their guidelines published in Japanese. (Robin Bennett is still looking for someone to proof for typos!)

■ Jill Fischer is our emissary to an NCHGR steering committee encouraging medical specialties to include genetics in their practice.

■ Rosalie Goldberg, Gladys Rosenthal and Laura Zajac will present our genetic counseling perspective to the multidisciplinary group writing clinical guidelines in New York.

Perspectives in Genetic Counseling is published quarterly by the National Society of Genetic Counselors, Inc. Editorial Staff:

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Send articles, reviews, correspondence to the Editor-in-Chief; address changes, subscription inquiries, advertisements and classifieds to the Executive Director. Contact any Editorial Board member with ideas or suggestions.

Publication date, next issue: June 15
Deadline for submissions: May 10

CPT Codes

Deb Lochner Doyle joined Wayne Grody, MD, and Mike Watson, PhD, of the American College of Medical Genetics as they testified before the American Medical Association CPT Panel, defending proposed descriptive codes for genetic counseling services. If accepted, these codes will be a major step towards reimbursement. The team represented the issues well; now we await the panel's response. Thanks to Deb, Barbara Bernhardt and all those who assisted with this strong effort.

NSGC Evolves

One of NSGC's strengths is our thoughtful and strategic approach to internal growth. We are considering the most responsible way to take advantage of electronic communication. Our marketing plan will lay a strong foundation for future growth.

Your Response Counts

You will soon receive our bi-annual Professional Status Survey. For the first time, we'll have data to evaluate longitudinal responses. As always, honest and complete responses from as many members as possible make the tool more useful. Survey results are used extensively—it's our strongest document defining who we are and what we do. Carve out some time to respond and be counted!

Thanks to all those genetic counselors creating and accepting opportunities to demonstrate our unique value—greater by far than merely the salary we earn.

Vickie Venne, MS
President

Sharing Thanks

Honoring our Community

In 1994, a most cherished honor was bestowed on me—the Natalie Weissberger Paul National Achievement Award. At the time, I could not find adequate words to express my acknowledgements and thanks. Not until October's Annual Education Conference, seeing more than 600 genetic counselors—many of whom I've taught, worked, collaborated or spoken with—did I suddenly know why I was so thankful, what I could not express as I held back tears last fall! I feel deeply proud of our genetic counseling community. I am grateful and in awe of the connections we create as we work to achieve common goals.

I feel awkward being honored for what I have given when, in truth, I've received so much. Maya Angelou's words echo my sentiments:

> *It is better to give than to receive....giving liberates the soul of the giver. The size and substance of the gift should be important to the recipient but not to the donor, save that the best thing one can give is that which is appreciated. The giver is as enriched as is the recipient, and more important, that intangible but very real psychic force of good in the world is increased.*

There is truly no greater honor than to be recognized by one's peers. But this honor deserves to be bestowed on all of us who have built and maintain our profession. I share this award with all those who provided the community in which I participate.

Beth Fine, MS
Chicago IL

NSGC Notes

Legislative Alert!

■ Call NSGC's Legislative Hotline—610-872-7608, #5—to learn more about the Kassebaum-Kennedy insurance reform bill, which includes genetic issues. The bill is expected to be heard on the floor in the next few weeks. Pick up your phone to educate yourself and then your legislators!

Committee Activities

■ The **Genetic Research Issues** Subcommittee of the Social Issues Committee is compiling a bibliography on genetic research issues.

■ The **Social Issues Committee** has completed a Position Paper on folic acid supplementation of food to lower risk for neural tube defects. The paper has been sent to the FDA and will be published in the *Journal of Genetic Counseling*.

■ Over 100 counselors responded to the recent search for mentors by the **Membership Committee**. Mentor pairings will be guided by responses to follow-up questionnaires, matching by specialty and region whenever possible.

Liaison Reports

■ The **NCHGR Task Force on Genetic Testing** has issued a working document on the Principles of Genetic Testing, to be discussed at their April 1st meeting. To receive a copy or offer comments, contact our Task Force representative, Katherine Schneider, 617-632-3480, EM: Katherine_Schneider@DFCI.harvard.edu.

■ Rosalie Goldberg reports that the February meeting of the **National Advisory Council for Human Genome Research** reviewed recent events involving genetic discrimination and health insurance. Dr. Francis Collins calls the consensus paper in *Science* 270:391-3 "a nice muscular document."

Celebrate Ourselves!

■ **Karen Greendale** is co-principal investigator of a grant awarded to the American College of Medical Genetics to develop clinical practice guidelines. Awarded by the New York State Health Department, the grant will address appropriate standards for genetic counseling/testing for breast cancer as well as for evaluating newborns with birth defects.

■ The data report written by **Liz Stierman**, *Birth Defects in California: 1983-1990*, won the 1995 Gold Award for Excellence in Public Health Communication in the specialty items category. The competition is sponsored by the National Public Health Information Coalition.

■ **Vickie Venne** was awarded a grant from the National Action Plan on Breast Cancer and NIH to determine if peer educators can successfully educate women with breast cancer about genetic risk factors. The two year project is being conducted with Reach to Recovery Volunteers from the American Cancer Society.

Ethics Consults Available

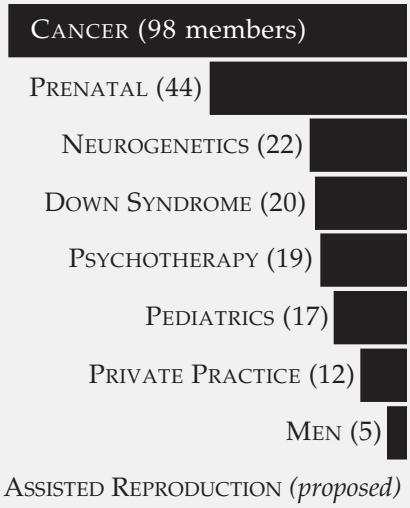
Contact any member of the Ethics Subcommittee for confidential consultations on ethical issues confronting you or your institution. Consults can clarify the role of the counselor as outlined by the Code of Ethics. This committee does not serve as a regulatory board or ombudsman.

Karen Eanet, Chair	410-706-3815	June Peters	301-933-9362
Sandra Peacock	713-781-1680	Robin Gold	313-493-6060
Beth Balkite	800-848-4436	Barbara Bernhardt	410-955-7894
Linda Nicholson	302-651-4234	Kevin Fitzgerald	708-216-4662

NSGC by Numbers

Special Interest Groups

Over 200 NSGC members have joined a Special Interest Group; several new groups have been proposed. For more information about a particular SIG, contact the person(s) listed at right.



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Cancer Risk Counseling: Aspirations and Admonitions

■ **A family's primary need may not be for susceptibility testing.** Information about risk factors, prevention guidelines and supportive counseling are an essential adjunct to testing and are especially critical for those with strong family histories who are not eligible for testing protocols.

■ **Empiric data for risk assessment must be applied carefully.** We must understand the limitations of different approaches, especially those that do not consider specific family history parameters or which combine empiric data with modeled analyses. For example, Claus *et al.* have published age-specific risks of breast and ovarian cancer based on family history.[1] Although widely used, these statistics overestimate risk once BRCA1 and BRCA2 mutations are excluded.

■ **Testing strategies should reflect an individual's family history.** Genetic analysis may be stepwise, reflecting the probability of finding specific mutations. For example, a family with breast cancer in a male may be initially evaluated for mutations in BRCA2, where affected males are more common.

E-mail/FAX Poll

Are You Ready for Clinical Testing?

Several commercial labs are offering BRCA1 testing as a clinical service, targeting patients at high risk because of family history and/or ethnic background. Should testing be available to all women or limited to research protocols?

☛ Contact Liz Stierman
FAX 213-380-7344
e-mail LStierman@aol.com

■ **Rapid screening tests may play a role in cancer susceptibility testing.** Although not a substitute for full sequencing, screening for a panel of common mutations may be a first step in high risk populations, such as Ashkenazi Jewish women with family histories of breast/ovarian cancer. In some cancers, such as the replication error phenotype in colon tumors, initial screening can determine whether further tests are needed.

■ **Counselors must understand the strengths and limitations of different testing approaches.** This task can be complicated—test sensitivity/specificity, genotype/phenotype correlations and the incidence of benign polymorphisms are not well established.

Delivering Services

■ **The demands for genetic testing and counseling for individuals of all risk levels must be met.** Some institutions triage clients according to risk; others have a hierarchy of services, including education or counseling provided in groups or individually by genetic counselors, nurses or other health care professionals.

■ **Informed consent protocols are paramount.** Informed consent is increasingly seen as an interactive, ongoing process rather than merely a document to be read and signed.

■ **We need more data on the psychosocial impact of cancer susceptibility testing.** We await information about short and long term effects of genetic counseling/testing on psychological well-being and medical decision making and a better understanding of what characterizes and motivates individuals to pursue testing.

■ **Insurance issues must be resolved.** There is great concern that insurance companies might use susceptibility testing to limit or deny coverage. Such discrimination may deter individuals not only from testing, but from seeking basic health care or learning about preventive measures. These public health implications are worrisome.

The Scientific Questions

In addition to examining the psychosocial impact of testing, a number of scientific questions need to be answered.

■ **How many major cancer susceptibility genes exist?** For instance, it is estimated that only 10% of breast cancers are due to germline mutations, most in the BRCA1 or BRCA2 genes.

■ **What are the most effective means of preventing, detecting and treating cancer in mutation carriers?** The role of tamoxifen and other chemoprevention agents, exogenous hormones, diet, alcohol, exercise and risk-reducing surgery in causing or preventing cancer need to be delineated.

■ **What are the relative weights of genetic and environmental factors in cancer?** Genetic counselors can be instrumental in developing clinical risk assessment tools based on emerging molecular and epidemiologic studies.

■ **How will we clinically apply a statistical probability of risk reduction?** This is especially challenging given that cancer is a disease of multifactorial origin and most cancers are not due to inherited predispositions.

The Road to the Future

As new susceptibility genes and mutations are identified and defined, testing will become simpler. We may then be able to test at-risk

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Positioning Ourselves: NSGC Devising Marketing Plan

organizations, by other health care professionals, and the public—those who will ultimately refer to and/or use our services."

This same rationale was used by the Board of Directors to identify four target audiences, listed in order of priority:

■ **Managed care.** Who are the decision makers spearheading the rapid changes in our evolving health care system and how can we effectively educate them?

■ **Medical specialties.** Which are most likely to interface with genetic counseling in the future?

■ **The public.** How can we ensure they understand our services and the value of genetic counseling?

■ **Potential students.** What can we do to attract excellence and diversity among those entering our field?

Research will Guide Plan

The marketing plan will consist of two phases. During the *strategic* phase, research will characterize the challenges we face by identifying who are making decisions, what they need to know about genetic counseling and the best time and manner to reach them.

Guided by this research, we will develop a strategy for the second phase—*implementation*.

Finding the Right Firm

After networking to find local marketing companies specializing in the needs of not-for-profit organizations, I narrowed the field to three. I supplied each with materials describing genetic counseling and NSGC, including our Strategic Plan Report, recent Annual Reports and our 1994 Professional Status Survey.

Each firm submitted an initial proposal describing its approach. They are now preparing final proposals, to be judged by a small ad hoc committee appointed by Vickie Venne.

Timeline

Key players from the company selected will attend the April Board of Directors meeting to begin their strategic work. They will conduct the bulk of their research by October, culminating with a report and recommendations. We hope to launch implementation by the end of 1996.

We may not know if the falling tree in the forest makes a sound, but with the right tools, we know genetic counseling's voice will be heard.

Marketing in Action

Following reports that members of health maintenance organizations were encountering difficulties in accessing genetic services, the New England Regional Genetics Group's Education Committee decided to focus their efforts on regional HMO medical directors.

Our project began with a written survey of 37 medical directors to determine how many HMOs included genetic services, what types of genetic testing were covered, who makes decisions about services covered and whether further information would be helpful.

We learned that HMOs continue to evolve. Many have merged, more genetic professionals have become in-network providers and awareness of the importance of genetic services is growing. Often the HMO medical directors make final decisions about services and funding. Most survey respondents requested further information; they will receive binders of genetic resources this summer.

Marisa Ladoulis, MS
NERGG Education Committee

Cancer, *continued from page 4*

individuals without first identifying a mutation in an affected relative. Difficulties will still remain: consider the challenges in interpreting a novel mutation or a "negative" test.

Currently, genetic analysis is a very complex process and still considered by most to be investigational. The previous points demonstrate that we are still determining the best way to identify mutations in cancer susceptibility genes. We remain a long way from understanding the epidemiological correlates.

Consumers and professionals are mobilizing to address these challenges before cancer susceptibility testing becomes the "standard of care." A sizeable grass roots force is lobbying for federal insurance protection. Also imperative is educating health professionals—particularly the gatekeepers in family practice, gynecology and oncology—about the responsible use of this new technology. How these issues are addressed will determine how to effectively triage genetic counseling and testing for high risk groups and to determine whether population screening will ever be appropriate.

References

1 Claus EB, Risch N, Thompson WD. Autosomal dominant inheritance of

The Prenatal and Childhood Testing Resolution...

■ *NSGC's newest resolution—addressing prenatal and childhood testing for adult-onset disorders—sparked a heated discussion at our Annual Education Conference. In the last issue, three experts commented on the resolution. This time, we present members' viewpoints.*

With the onslaught of new tests, genetic counselors may be increasingly challenged to serve as gatekeepers. Are you comfortable offering prenatal diagnosis for a gene predisposing to homosexuality? Would you support couples seeking prenatal testing for genes that contribute to intelligence? In either case, parents may feel they are acting benevolently in the best interest of their future child—to minimize social stigmatization or potential "suffering." Even after genetic counseling such couples are unlikely to alter their underlying judgments that heterosexuality and higher intelligence are superior traits in our society. We have a social responsibility to minimize such divisiveness.

As genetic counselors, we need to reconsider a broad-sweeping interpretation of nondirectiveness that suggests anything goes as long as the woman or couple has been counseled. In an effort not to influence reproductive decision making, some genetic counselors have adopted a hands-off model that may not be justifiable.

We have an obligation to protect children. Parents most often make decisions in their children's best interests. Sometimes they do not. This is one reason why vaccinations are mandatory.

Parents who desire testing of their children may unintentionally raise or treat their children differently based on their DNA status. Positive DNA test results in childhood may render a child uninsurable as an adult. Parents fixated on relieving their own anxiety and uncertainty may minimize these and other risks presented during counseling. Learning of these potential harms is unlikely to alter the long-term outcome. What parents intentionally raise their children differently?

Once they reach adulthood, children may choose not to know their future genetic destiny. This choice will have been taken from them if their parents' wishes are granted. There is a paucity of data to guide us in determining when an adolescent is "ready" to undergo testing. Despite this, the decision should be left to them as adults rather than to their well-meaning parents.

The NSGC position statement should not support the testing of minors for adult-onset conditions.

Barbara Bowles Biesecker, MS
Bethesda MD

The discussion about NSGC's Childhood Testing Resolution ultimately raises the issue of patient autonomy. If we as genetic counselors are willing to give our patients full autonomy, there is essentially no conflict with our resolution. If, however, we feel that counselors should be able to limit patient autonomy based on ethical principles—or indeed, if this is expected of us—then we must clearly define where those limits are. Of course, we could decide that situations such as childhood testing be handled on an individual basis or by an ethics committee or IRB. The decision is ours to make.

Pamela Cohen, MS
Charlestown MA

My main problems with the resolution concern timing and form rather than content. The resolution arrived just days before the Minneapolis meeting, printed in such small type that I frankly had trouble reading it. A resolution should be able to be stated in one or two sentences; the justification and explicitly detailed explanations would be better served in another format.

The first draft published in *Perspectives* was considerably shorter and less cumbersome. I find it amazing that two responses to that first draft provoked such major revisions. The final draft was changed enough to warrant a full review by the membership, not a take-it-or-leave-it vote.

Finally, in my opinion, if an adequate percentage of members (a quorum) do not vote on a resolution, it cannot be considered "resolved."

Susan A. Demsey, MS
Bellflower CA

Though NSGC resolutions should be distinct from those of our MD colleagues, this one is more than distinct, its concern for potential harms to children is fundamentally different from the ASHG counterpart. More importantly, the resolution diverges from past NSGC policy on genetic testing. It says that such testing should be offered—despite the dearth of research data on the psychosocial impact of presymptomatic genetic testing in children.

In the past, NSGC strongly opposed genetic testing programs until psychosocial data became available. In its position statements on genetic screening and cystic fibrosis, NSGC stipulated that genetic tests should be

...Members Express their Views

assimilated into clinical practice in the following manner: test outcomes must first be studied to determine what (if any) harms ensue before the test is offered on a more widespread basis.

The current resolution does not follow suit. It states that families "...should be made aware of clinically available testing technologies," and "until more data is gathered on the impact of this type of testing, extreme caution should be taken regarding the use of such tests." The distinction is not simply semantic, but a new message: counselors should offer these tests although the implications have not yet been examined. Why are we now willing to support presymptomatic diagnosis of children (even in the absence of therapeutic benefit) when pilot studies have barely begun?

Based on the NSGC's proud history of pulling in the reins on genetic testing, we could have called for a moratorium on presymptomatic testing of children outside of research protocols. Such protocols, incorporating genetic counseling and psychological services, would enable us to monitor and support families who desire testing.

One argument for testing is that we are obligated to apprise parents of all available tests to remain nondirective and non-paternalistic. But our past position statements and resolutions have been guided by another tenet of genetic counseling: nonmaleficence.

Limiting childhood testing to the confines of pilot studies until potential harms are clarified is certainly consistent with our profession's goals. Our Code of Ethics states our responsibility is to facilitate informed decision making by "...providing or illuminating the necessary facts and clarifying the alternatives and anticipated consequences." We are quite limited in our ability to fulfill these obligations when the consequences are unknown or only hypothesized.

Elinor Langfelder, MS
New York NY

I am glad the content of the resolution remains open for debate. Presentations and discourse at the ASHG and NSGC meetings helped me more critically evaluate the proposed resolution and to offer these comments and suggestions:

Address prenatal and childhood testing in separate documents. The issues surrounding childhood testing and prenatal testing are related, but distinct, given that for many the status of a fetus and a child is not the same.

Don't confuse nondirectiveness with "doing what the patient wants." Genetic counselors are trained to explore with patients the underlying forces behind their questions; not immediately jump through hoops and arrange for testing. I propose the document be reworked to support client-centered genetic counseling and to emphasize the evils of fulfilling parents' requests simply because the technology is available and they are able to pay.

Clarify "assent" and "informed consent." Statement #2 is flawed: a parent may never give informed consent for a child; an individual may only give consent for him- or herself. I propose it be stated that childhood testing may only be done when the parent has given consent *and* the child has given informed assent for genetic testing, not just a blood draw.

Review the approval process. It would have been helpful to read pros and cons regarding the resolution from those actively involved in the testing debate. I propose that future resolutions be presented to members with reviews in *Perspectives* written by people not involved in developing the resolution.

The timing of this vote prior to to the Annual Education Conference was misguided—the meeting provided critical information and opportunities for discussion. Consequently, I support a re-vote on the proposed resolution.

Kathy Peters, MS
Bethesda MD

Previous resolutions state our beliefs clearly and succinctly, as in this example: "The NSGC, as an organization, publicly supports a woman's right to reproductive freedom, including her right to prenatal diagnosis and access to safe and legal abortion." The use of non-technical language helps communicate our views to those outside our profession, such as reporters.

The current Childhood Testing Resolution goes much beyond these limits—I feel its thorough exploration of the issues should be reclassified as a Position Paper. If we truly want a resolution on this topic, let's condense our conclusions to one or two simple statements understandable by all.

Liz Stierman, MS
Los Angeles CA

■ *The Social Issues Committee and the Resolution Working Group appreciate this thoughtful feedback as they consider how to proceed. Look*

✍ ✍ ✍ Letters to the Editor ✍ ✍ ✍

Scrambling for Jobs

In light of changes due to managed care, graduate students looking for jobs and the approval of new graduate programs, I share with you the following:

Years ago I was at Yankee Stadium. The large crowd stayed until the end of the exciting game and then all exited the stadium at once. To exit, the crowd had to go down a series of escalators—down the escalator, turn right, down the next escalator, turn right, down the next escalator, etcetera.

Given human idiosyncrasies, with each descending level the crowd became more and more backed up, and there was less room on the platforms. Half way down one of these completely full, long escalators, I noticed there was barely enough room on the platform below to receive arriving passengers. As I was lowered further to my destination I saw there would be a spot for me on the platform.

Then it got increasingly tight. We packed in, we felt the squeeze, the pressure. It was clear there simply was no more room. But there were plenty of eager people already descending the filled escalator, coming at us one after another. Not everyone noticed the problem at first; people above kept entering the escalator.

Eventually the awareness traveled upwards but it was too late for many. Anxiousness gave way to panic, which gave way to screaming and eventually jumping. There were injuries.

Graduate program directors please take note.

Seth Marcus, MS
Park Ridge IL

A Salary Success Story

A compensation study was performed at our private non-profit midwestern hospital in 1994. We responded to a lengthy questionnaire evaluating the complexity of our daily activities in eight areas: knowledge, skill, work complexity, contact with others, property protection/use, work leadership, working environment and patient welfare. As a result of the study, genetic counselors were assigned to a job grade with the salary range of $25,900-$39,000 (comparable to the previous range, in place since 1987).

With our administrative director's support, we decided to appeal the job grade assignment. We submitted a written appeal, further characterizing and clarifying our knowledge, work complexity and patient welfare.

To document "job knowledge," we described entry requirements for a Master's genetic counseling program and summarized course work and clinical training in a typical program. To show "work complexity," we described the depth of our scientific knowledge, demands placed upon us to remain current, our ability to interpret lab reports and translate complex medical information for laypersons. Addressing "patient welfare," we discussed ethical and legal issues that could arise in counseling sessions and the impact of genetic information on patients' major life decisions.

We raised two salary-related problems: not being able to recruit top candidates due to salary limitations and limited salary growth for counselors who had reached the top of the pay scale. NSGC's 1992 and 1994 Professional Status Surveys illustrated the inequity between our hospital's range and regional/national salary trends.

In Fall 1995, we were notified our job grade had been upgraded by two levels, with a revised salary range of $30,500-$46,900!

We would be happy to assist other genetic counselors engaged in similar battles. Please feel free to contact us at 815-969-5069.

Suzie Stilwell, MS
Gina Morley, MS
Luna Okada, MS
Kristin Kruger Sanden, MS
Rockford IL

If I Can Do It, So Can You!

Recently at ASHG, Dr. Francis Collins mentioned how important it was for us all to become politically active and express our views to our elected officials. I had heard calls to action many times before, but for some reason, this time I decided to respond. It happened I was going to Washington DC two weeks later. I decided genetic discrimination in insurance was an issue worth discussing with my elected officials.

I called my senators' Kansas City offices, where I was referred to the health care legislative assistants in Washington. I left voice mail messages, asking to set up meetings when I would be in town. To my amazement, nothing happened. (In the back of my mind, I never expected to get an appointment, but I did expect phone calls saying their schedules were too busy.) A week before my trip, I left more messages, this time with "attitude" in my voice. I received return calls within 24 hours.

One of my state senators is Bob Dole, Republican majority leader

and the Republican frontrunner for president. To my surprise, I was offered an appointment with him as well as his health care assistant. Kansas' other senator is Nancy Kassebaum. Although not running for president, she chairs a powerful committee responsible for health care-related issues. I was scheduled to see one of her three health care assistants, who happens to be a physician.

It was only three days before I was to leave for Washington, and now I was nervous. Not really expecting to get appointments, I certainly wasn't prepared to talk to anyone! To gather information so I could present the issues intelligently and succinctly, I made phone calls—to my professional organizations, to colleagues, to NCHGR's legislative expert. I left for Washington armed with pages of material to prepare for my meetings.

I discovered Senator Kassebaum was lead author of an insurance reform bill directly addressing the issues I felt were important. In my visit, I supported and thanked the Senator for her efforts. I expressed my opinion that the bill was missing elements dealing with genetic information. Senator Kassebaum's assistant assured me this information would be included in the bill.

After I met with the legislative assistants and senators, you would have thought I had dinner with the President—it is an exhilarating and incredibly rewarding feeling!

If I can do it, so can you. Get to know your elected representatives; let them get to know you. The process is not difficult but the payback is immense.

Amy Strauss Tranin, RN, MS, OCN
Kansas City MO

CYBERGENES

Let's Get Caught in the Web

It's time for us to discuss the Internet's fastest growing, most celebrated segment—the *world wide web* (WWW). The web is a hypertext system, one in which related documents or sites are linked to one another. Users can access these links to move between different parts of the same document or to travel to different sites. If you can move a mouse and click, you can navigate the web. It's an easy way to research any subject in a short time, merely by clicking—I predict this is how most of us will conduct research from now on.

SET YOUR SITES ON THE WEB

A *web site* is simply a document that can be accessed by anyone with an Internet connection. Web sites can contain text, graphics, sound and movies about a specific topic. There are thousands of web sites with information about every topic you can imagine—the sciences, arts, sports, politics, government. Individuals, like my computer geek brother-in-law, can post their wives' ultrasound pictures on their own web sites.

Each web site, often called a *web page*, has its own address. Known as the *URL* (uniform resource locator), the address always appears on the top of your screen. You probably have seen this type of address—many companies now list their web site URL in commercials, for example, http://www.toyota.com. The software you use to access the Internet will have a place for you to type in the URL for your desired destination.

A HOMEPAGE IN THE HOOD

The *homepage* is the introductory document for a given web site. Think of a web site as a folder containing a variety of documents—the homepage is its cover, describing what's inside. Most homepages also display related information located at other web sites, with links to take you directly there.

WHAT EXACTLY IS A LINK?

No, it is not a big spicy sausage from the South. A link is a way to move from one document to another simply by clicking on a highlighted word. Text about linked topics is underlined or shown in a different color, for instance, in blue. When you click on one of these highlighted blue words, you automatically jump to a new page about that topic. The new page can be within the original site or in an entirely different location.

It may sound complicated but it's really quite simple. You can learn the basics in about five minutes on-line. The WWW is intuitive; there are no commands to memorize—all you have to do is point and click. A good place to start is the Genetics Education Center homepage designed by Debra Collins at the University of Kansas. This great home page has many helpful links. You can access this site at: http://kumchttp.mc.ukans.edu/instruction/medicine/genetics/homepage.html.

I wish everyone good luck and remember... always wear a life jacket when surfing the web.

Steven Keiles
Steven.Keiles@kp.org

Bulletin Board

Ethics Cases Wanted

The Ethics Subcommittee seeks cases for the "Difficult Dilemmas" Workshop at the 1996 Annual Education Conference—the theme is "Dealing with Other Disciplines." Cases may include dilemmas with labs, Children's Protective Services referrals and differences in care philosophies between counselors and other health professionals. Those who submit cases do not have to be present at the workshop, and as always, confidentiality will be maintained. Therefore, no names or other identifying information should be provided, other than a contact person to clarify questions.

☛ Fax cases to Karen Eanet, 410-706-4059, or contact any committee member *(see page 3)*.

Visiting Fellowships in Biomedical Ethics

Stanford University's Program in Genomics, Ethics and Society is sponsoring visiting fellowships of one to three academic quarters, supporting research and participation in the PGES Working Group. Fellows—who generally hold postgraduate degrees in genetics or related fields—receive a stipend, office space and some research support.

☛ For more information, contact: Henry Greely, 415-723-2517, EM: hgreely@leland.stanford.edu or Barbara Koenig, 415-725-6103, EM: MR.BAK@forsythe.stanford.edu.

New Deafness Manual

An Introduction to Deafness: A Manual for Genetic Counselors by Jamie Israel is available to genetic counselors for approximately $15 (exact cost is yet undetermined). Supported in part by an NSGC Special Projects Award, the guide has already been sent to genetic counseling training programs.

☛ To order, contact Kathleen S. Arnos, PhD, at Gallaudet University, 202-651-5258 V/TTY.

☛ For more information about the project, contact Jamie Israel at her new number, 414-942-9946.

New Special Interest Group: Down Syndrome

Down Syndrome officially joins the ranks of NSGC Special Interest Groups. Are you interested in participating? Do you have ideas for the group's format?

☛ Contact Cam Knutson Brasington, 704-355-3159.

Clinical Supervisors' SIG

Are you one of those exceptional people involved in the challenging but oh-so-rewarding task of training student genetic counselors at their clinical sites? Want to share experiences and tips with others like you around the country? Perhaps it's time to start a Clinical Supervisors Special Interest Group.

☛ Contact Liz Stierman, 213-380-5362, EM: LStierman@aol.com.

Survey Coming

Full NSGC members will soon receive questionnaires about screening practices for cystic fibrosis and Canavan disease—please complete and return them promptly. The survey is part of a thesis project being conducted by Brandeis genetic counseling students Noelle Myles Bodkin and Stephanie Snow.

Upcoming Meetings

April 12-13	"Conversations: Personal, Professional & Ethical Challenges in the Treatment of Breast Cancer," from the Center for Biomedical Ethics, Minneapolis MN. Contact: 612-626-9756.
April 17-19	Nature Genetics & Canadian Genetics Diseases Network Conference: "Genetic Susceptibility & Complex Traits," Vancouver, Canada. Contact: 212-726-9281.
April 24-27	American Cleft Palate-Craniofacial Association Meeting, San Diego CA. Contact: 412-481-1376.
April 28-30	Human Teratogens Course by the Harvard Med School & Mass General Hospital, Boston MA. Contact: 617-432-1525.
May 3-5	Univ of Pittsburgh/NSGC Genetics Board Review Course, Pittsburgh PA. Contact: 610-872-7608, EM: beansgc@aol.com, or 412-624-9951, EM: bgettig@helix.hgen.pitt.edu.
May 17-19	Genetics Review Course, Baylor College of Medicine, Houston TX. Contact: 713-798-6020.
June 13-15	TEXGENE Annual Conference: "Genetics and Managed Care: Probing the Present for Future Solutions," Austin TX. Contact: Trudy Jones, 512-458-7700.
July 14-19	Midwest Intensive Bioethics Course, from the Center for Biomedical Ethics, Minneapolis MN. Contact: 612-626-9756.

PLANNING FOR 1997

Yes, we said 1997! It's not too early to think about NSGC's 16th Annual Education Conference, to be held October 24-28, 1997 in Baltimore. Co-chairs Barbara Pettersen and Cindy Soliday are looking forward to the challenge of planning the event and encourage anyone interested in assisting to get in touch. Working on the conference can be a great way to become more involved.

☛ Barb Pettersen, 408-972-3311, EM: Barbara.Pettersen@ncal.kaiperm.org; Cindy Soliday, 510-795-9478, EM: CESoliday@aol.com.

GENETIC TESTING STORIES SOUGHT

The Task Force on Genetic Testing, created by the Working Group on Ethical, Legal & Social Implications of Human Genome Research, is eager to learn about specific experiences —both good and bad—of genetic counselors, nurses, consumers or others who order genetic tests and/or receive results. The information will assist the Task Force in developing principles to ensure genetic tests are safe, effective and provided in labs of assured quality.

They are especially interested in informed consent; communicating/counseling about lab test results, including reporting speed, patient satisfaction; confidentiality; conflicts of interest; and the role of institutional review boards in deciding test use.

To preserve confidentiality, don't cite names of labs, organizations or specific individuals, but do include the condition involved and the reason for testing.

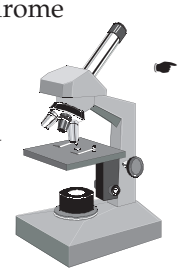
☛ Neil A. Holtzman, Chair; Task Force on Genetic Testing; 550 N. Broadway, Suite 511; Baltimore MD 21205; 410-955-7894; EM: holtzman@welchlink.welch.jhu.edu.

Research Network

COMPLEX DISORDERS COLLECTION

The National Institute of General Medical Sciences Human Genetic Mutant Cell Repository is requesting blood or lymphoblastoid cell cultures from probands with well-documented phenotypes representing a variety of familial complex genetic disorders:

- asthma
- atherosclerosis
- attention deficit disorder
- autism
- cancers (familial): breast, prostate, colon, pancreatic, melanoma
- cataracts
- cleft lip and palate
- congenital heart disease
- Crohn disease & other inflammatory bowel disease
- dyslexia
- epilepsy
- fetal alcohol syndrome
- glaucoma
- hearing loss (nonsyndromic)
- hypertension
- long QT syndrome
- lupus erythematosus
- macular degeneration
- migraine
- morbid obesity
- multiple sclerosis
- neural tube defects
- osteoporosis
- Parkinsonism
- psoriasis
- rheumatoid arthritis
- Tourette syndrome



To arrange to submit specimens, contact:

NIGMS Human Genetic Mutant Cell Repository
Coriell Cell Repositories
401 Haddon Avenue
Camden NJ 08103

Looking Forward to San Francisco

15th Annual Education CONFERENCE
nsgc

October 26-29 are the dates of our upcoming Annual Education Conference at the Grand Hyatt Hotel on Union Square in San Francisco. Genetic counselors' diversifying professional needs and workplace settings will be explored, along with recent advances in genetics and our changing health care system. Once again, practice-based symposia will bring together those working in the same specialties to share resources and build professional alliances.

Preceding the conference is a 1 ½ day short course, "Unlocking the Secrets of Neurogenetics," a clinical look at ataxias, phacomatoses, neuromuscular disorders, muscular dystrophies, Alzheimer disease and familial dementias. The course will also review neurologic terminology, neuroanatomy and diagnostic procedures.

Survival Skills for Genetic Counselors

Guidelines for Writing Grant Proposals

■ *Perspectives proudly launches a regular new feature—**Survival Skills for Genetic Counselors**—where our colleagues share their expertise in topics you probably didn't cover in graduate school. This is the first of two parts on grantwriting.*

Genetic counselors have a growing interest in clinical research and grantwriting skills, demonstrated by the outstanding response to the Jane Engelberg Memorial Fellowship Grantsmanship Seminar, sponsored by a grant by the Engelberg Foundation to the NSGC. We were extremely pleased to receive 58 applications for the training session, which took place on February 8 and 9. The next issue of *Perspectives* will include a full report on the seminar's outcome.

Many applicants submitted exciting research ideas, presented as responses of 300 words or less to three questions: "What is the goal of your project?" "How will you accomplish this goal?" and "How will you measure the effectiveness of your project?" Our underlying criterion when reviewing applications was to choose those with the highest potential to submit fundable proposals to granting agencies in the near future. The quality of the applications made it very difficult for the advisory board to limit attendance to 22 genetic counselors based on available funds.

As we reviewed the applications and formulated our selection criteria, the advisory board developed these seven suggestions for writing grant proposals:

■ **A great idea isn't enough.** Sound and often innovative ideas must be converted into researchable projects.

■ **Know and cite relevant literature.** Validate the scientific content of your proposal with citations supporting factual statements. Conduct a literature search, document the need for the proposed project and include references.

■ **Research methodology must adequately address the project's goals.** As you design your study, make use of expert advice on recruiting subjects, sampling, designing questionnaires, using validated instruments, piloting materials, differentiating between qualitative and quantitative approaches, analyzing data and testing hypotheses. Seek assistance with designing and working with a research team. Become familiar with the role of consultants and advisors.

■ **Make plans for evaluating the project's outcome.** Proposals should include clear and relevant procedures for measuring your project's success and impact.

■ **Adhere to professional and institutional research guidelines.** Proposals should document the required protection for human subjects. Counselors must be especially sensitive to potential coercion, testing of children for adult-onset disorders, inclusion of minors in research protocols and the unauthorized use of membership lists from voluntary disease organizations. To address these issues, counselors should have a thorough grounding in the theory of informed consent and the role of institutional review boards.

■ **Don't overlook submission guidelines!** Make sure you follow instructions relating to formatting, page length, deadlines for submitting applications and other guidelines.

■ **Writing and presentation skills are critical.** Make sure paragraphs are focused with clear transitions; avoid long or cumbersome sentences. Be careful not to overuse passive voice or long adjectival/noun phrases; check for proper grammar.

We encourage genetic counselors interested in clinical research to write proposals. We wish them success with their research ideas.

The Jane Engelberg Memorial Fellowship Advisory Board:

Audrey Heimler, MS, Chair
Bonnie J. Baty, MS
Barbara A. Bernhardt, MS
Barbara Bowles Biesecker, MS

NSGC Deadlines

April 1	Nominations for the Natalie Weissberger Paul National Achievement and Regional Leadership Awards
April 1	Nominations for NSGC elected positions
April 26	Call for Abstracts, 15th Annual Education Conference
May 1	Jane Engelberg Memorial Fellowship Proposals
May 15	Applications for Special Projects Fund
	Contact the Executive Office for more information or applications: call 610-872-7608, #8; FAX 610-872-1192; EM: beansgc@aol.com.

FAX/E-mail poll

For Guidance and Support: More Thanks for Mentors

■ My program director at Rutgers was **Marion Rivas**. She created a most unusual atmosphere (for 1972) for a graduate program: one of co-operation and non-competitiveness. Students helped each other with weak areas, studied together and by graduation we were treated by our instructors as colleagues. This atmosphere of mutual respect made a deep impression on me and has been a key feature in my understanding of the counseling relationship, thanks to Marion's modeling.

Bonnie Baty, MS
Salt Lake City UT

■ **Dorothy Wertz** supervised my research project on ethics and genetics while I was a student at Brandeis. She opened my eyes to key issues and controversies in genetics today and help guide me through the world of quantitative and qualitative research methods. We spent countless hours discussing my project and my career options as a counselor interested in ethical issues. Thank you Dorothy for inspiring me to pursue my research interests and for supporting me as a student and now a colleague.

Pamela Cohen, MS
Charlestown MA

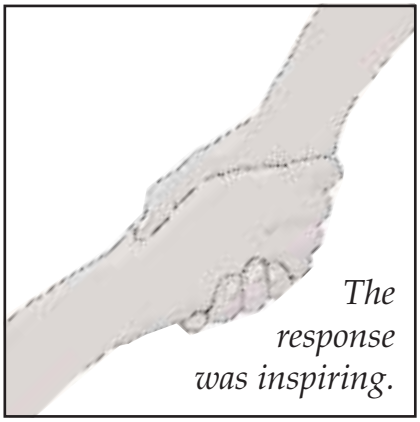
■ I will be forever thankful to four mentors: **Ann C.M. Smith** provided an impressive role model as a genetic counselor and on the NSGC and ABGC Boards. Her commitment, intelligence, good nature and constant support give me an ideal to work towards. We have become dear friends and compatible collaborators!

The late **Beverly R. Rollnick** was a mentor in my roles with NSGC. She taught me to look to the future after learning from the past. Beverly had a terrific sense of the "big picture" and held lofty goals for our profession. I derive great satisfaction knowing how proud she'd be of NSGC today!

Mark Lubinsky taught me to think critically, follow my heart and intuition and be creative in all aspects of my career. Mark was an excellent role model for co-counseling, teamwork and exploring academic interests within a clinical setting. Our work together was some of the most fulfilling of my career.

Eugene Pergament has been my most influential mentor. He taught me all problems can be solved while focusing on patient care as an ideal. His integrity in clinical research and genetic counseling should be a model for all genetics professionals. His encouragement and support have helped me reach my goals.

Beth Fine, MS
Chicago IL



■ It's a little difficult to think of **Sandra Davenport** as my mentor, as over the years she has also become a colleague and friend. We first worked together on a "Deaf-Blind Project" which fairly quickly became a CHARGE association project. Sandy is a straightforward person who gives and is comfortable receiving constructive criticism, a wonderful way to learn and grow. Now we are in different cities, but continue to work closely to help families whose children have CHARGE.

Meg Hefner, MS
St. Louis MO

■ When I became interested in this profession, I called a genetic counselor. Little did I know how lucky I was there was one in Binghamton NY in 1983. She gave more than just information—she provided support, encouragement and set me on the right path. She demonstrated the many different ways an individual can work in our field. For all this and more, I say thank you to my mentor, colleague and friend, **Luba Djurdjinovic**.

Steven Keiles, MS
Los Angeles CA

■ My mentor memory is of **Jessica Davis**. I was trying to juggle classes at Sarah Lawrence, a part time job, two children and a marriage that was in trouble. I was interviewed by a group of people evaluating the Sarah Lawrence program. Afterward Jessie came up to me; I can't recall her exact words but the message that came across was "You seem to be juggling a lot on your plate and working really hard. I'm here to help you and I believe in you." It meant so much to me that she cared at a time when I really felt that no one cared very much at all.

Jodi Rucquoi, MS
New Haven CT

■ The variety of positions I have held over 18 years allowed me to learn from, commiserate with and enjoy many colleagues, but I consider four to be special mentors: **Myrna Ben-Yishay**, who gently taught me it is okay to make mistakes and forgive myself; **Rosalie Goldberg**, who is so creative; **Elsa Reich**, who helped me define an incredible standard of excellence; and **Pat Ward**, who remained calm through all my passion and helped me mellow. There are many others. What a joy to be colleague to such marvelous professionals.

Vickie Venne, MS
Salt Lake City UT

Resources

■ Study Guide ■

Medical Genetics: Overview & Study Guide

by Janice L. Berliner, MS. Published by Janice Berliner, 34 Webster Drive, Berkeley Heights NJ 07922, 908-771-5582; 2nd Edition, 1995; $63.95, volume discounts available.

It's three months before the ABGC exams. Study time. You're pulling out all those old human genetics class notes and molecular genetics textbooks, each a sea of details you must wade through to reach your goal—passing the boards!! Fortunately, you find your unopened copy of *Medical Genetics Overview and Study Guide* by genetic counselor Janice Berliner. You breathe a sigh of relief as you begin to organize your review using this practical and concise manual as the basis for your productive study efforts.

The study guide is self-published in a soft cover spiral notebook. Organized into twelve chapters of discrete subject areas, it is simple to skip around to items of particular interest or concern, especially with the help of the comprehensive index.

Chapters are easy to read and thorough. Although there are few visual aids in the guide, the chapters are of attention-span length. Ms. Berliner deftly summarizes a vast array of topics in a clear and succinct format. Spot checks of facts and figures did not reveal any obvious errors.

Readers educated in genetics should have few problems reviewing the material. However, this is a guide in the true sense. In subject areas in which I had little previous exposure, I needed other references to increase my understanding.

Practice questions were the only major component I found lacking. Providing a few questions at the end of each chapter would be beneficial to many readers. In addition, the study guide is very text heavy and dense, making for dry reading at times. Since human genetics is an exciting field (part of the reason we chose this career), a more dynamic presentation of the material would be helpful.

...It's three months after the board exam. Long hours of LOD scores and metabolic pathways have become distant memories. With the guidance of your *Medical Genetics Overview*, you were confidently prepared for the big day. Your hard work has paid off as soon as you open that official letter in your mailbox...

Melanie Andrews-Casal, MS
Austin TX

■ Books ■

Genetics and Human Health: A Journey Within

By Faith Hickman Brynie. Published by the Millbrook Press, Brookfield CT, 1995, 128 pages.

According to the author, this relatively short text on genetics is intended for young adults, ages 12 and older. This is a wonderful age group to target since an early interest in genetics may lead to a career in the field.

Brynie takes the reader on a journey, each chapter representing a leg of the trip. We travel to the Olympic sports arena to learn about Marfan syndrome and Flo Hyman and then to Eastern Europe to meet Gregor Mendel. The journey metaphor works well and personalizes the information.

An interesting chapter on Mendel incorporates historical lore about his life and personality which makes him come alive. This is also true of chapters on Watson and Crick and on Huntington disease, which includes a personalized look at Nancy Wexler and her involvement in the hunt for the disease gene.

Unfortunately, the book's descriptive science is pretty dry—more illustrations, photographs or colorful diagrams would help. Explanations of inheritance patterns, chromosomes and genes would be very difficult for the younger reader; it is hard to imagine a 12 year old reading this book. However, the section on protein synthesis was quite good. The analogy of sending a message by Morse code to someone who does not speak your language was descriptive and easy to follow.

The author includes thought-provoking ethical questions about predictive testing and insurability. Gene therapy and prenatal diagnosis are also touched on briefly. A helpful section at the end lists books, articles and pamphlets for those wanting more information; it includes the NSGC's address.

Although it may not keep the full attention of the younger reader, hopefully, the role models and interesting characters presented in this book will spark readers' imaginations and send them on a more in-depth "journey" into the genetics field.

Patti Robbins-Furman, MPH

Resources *continue on page 16*

national society
of genetic
counselors, inc.

nsgc

233 CANTERBURY DRIVE • WALLINGFORD PA 19086-6617

Resources, *continued*

SHATTERED DREAMS—LONELY CHOICES: BIRTHPARENTS OF BABIES WITH DISABILITIES TALK ABOUT ADOPTION

By Joanne Finnegan.
Published by Bergin & Garvey, Westport CT, 1993, 184 pages, $22.95.

Since resources for parents considering adopting out a child with a disability are scarce, this book is an important one for parents and professionals. Ms. Finnegan chose adoption for her son with Down syndrome; this book is her "gift" to parents grappling with this difficult decision. She hopes it will fill part of the void of information on this subject and support positive attitudes about adoption.

The author's hopes have been well-realized. The book is nicely formatted with a blend of quotes, information, touching poems and an informative appendix. It does not preach what is right, wrong or normal; instead, it gives a sense of what many parents have felt before, during and after the adoption process.

Each chapter begins with an insightful quote from a parent who has considered adoption and ends with a poem. In a section called "Parents Talk" (usually as long as the chapter itself), parents give candid reactions to the chapter's topic. The last part of each chapter, called "Note to New Parents," offers concise advice.

The suggestions for parents considering adoption are helpful, things most parents in this situation would not have thought of on their own. The end of the book has a useful appendix which lists available resources including books on a variety of disabilities, grief/loss information and national organizations.

Unfortunately, the author apparently did not consider genetic counselors as a resource—they are not mentioned as professionals for parents to contact during their search for information, nor are they listed in the appendix. Most people facing this type of decision in such a limited time period would not have the energy or resources to investigate the many sources of information suggested by the author.

It appears that adoption is not viewed as a "real" option by many professionals. This book is a big step toward validating this alternative and could be used in training programs for genetic counselors, social workers and physicians to help change their perceptions.

Amy Vance, MS
San Francisco CA

■ Support Group ■

OVARIAN CANCER RISK

Gilda Radner Familial Ovarian Cancer Registry
Roswell Park Cancer Institute
Elm and Carlton Streets
Buffalo NY 14263
1-800-OVARIAN

A telephone helpline offering peer support to high risk women. Helpline volunteers are high risk women who share personal experiences about health care options and screening methods.