PERSPECTIVES
in genetic counseling

Volume 26 Number 4 Winter 2004

national society
of genetic
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the leading voice, authority and advocate for the genetic counseling profession

www.nsgc.org

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NSGC Publications at Work!

Order Your Hard Copy of *JGC*

In response to requests to retain the hard copy option of the *Journal of Genetic Counseling*, our publisher, Springer, will offer print copies of the year's entire volume.

Here's how it works:

- NSGC first will collect the $75 fee from each member who wants a print copy, and
- we will place a group order for the volume of the year just completed.

Other fees apply for non-members.

Currently we are taking orders for hard copies of Volume 13 (2004). **Orders are due by Tuesday, February 15, 2005 – no exceptions!** You will receive your bound volume of the *Journal* directly from Springer. Payment can be made online, by check or by credit card.

Search *PGC* All the Way to Volume 1

Want to see what genetic counselors were concerned about 10, 15, even 20 years ago? *Perspectives* now has been archived back to the first issue in 1979. A special search tool on the *PGC* webpage allows you to find topics by keyword. So jump online to research information for your practice, or just browse for fun through the living history of our profession! ❖

☞ www.nsgc.org/members/perspectives

A Growing Patient Population: Understanding the Military

Courtney Sebold, MS

In graduate school we are all trained in the importance of multiculturalism. However, not all cultures are defined by national borders or ancestry. As a genetic counselor who works in a military health setting and who is part of a military family, I have learned that a culture also can be defined by career choices. The military culture is unique and presents challenges for the genetic counselor as well as the family.

Active Duty

There are hundreds of military installations spread across the United States. There are approximately 1.5 million active duty men and women among the services. There also are 1.1 million reservists, the majority of whom live in the US, and ...to page 5

President's Beat

It is with great pleasure that I write my first column as your NSGC President. I enjoyed the opportunity to see old friends and meet new faces at our recent Annual Education Conference (AEC), and I am excited about the possibilities for NSGC as we celebrate our 25th year.

Strategic Plan

NSGC continues to make strides in achieving our current strategic plan. **Dawn Allain** and I moderated a discussion at the AEC, with over 75 attendees, where we reviewed the goals of the strategic plan and some of its specific points. We hope to host further discussions at the 2005 regional meetings.

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

Next issue ***March 15***
Submission deadline ***February 11***

Items to Watch

Official membership voting on changes to the code of ethics has just ended. Look for results soon at www.nsgc.org. Membership voting on the definition of genetic counseling, developed by **Robin Bennett** and committee members, will soon be available. Also, starting in January, expect some changes in the *Journal of Genetic Counseling* as our newly medline-indexed journal gets a new look and goes to a larger page size. For those of you craving the paperbound journal, make sure you sign up for the new year-end option on your dues form.

Kelly Ormond, MS

Jumping Right In

In my first months in office, I presented testimony on genetic discrimination and reimbursement of genetic services at the Sectretary's Advisory Committee on Genetics Health and Society. Other genetic counselor presenters included **Don Hadley** and **Jeff Shaw** (on an invited panel) and **Andy Faucett** representing the American Board of Genetic Counseling. I also was present at an event to thank **Representative Don Moffitt** for his assistance in the passage of the genetic counseling licensure act in Illinois. I was the NSGC representative for a National Human Genome Research Institute working group developing educational materials for a variety of genetics professions. Other Board members and I conducted meetings with American Society of Human Genetics, American College of Medical Genetics, Canadian Association of Genetic Counselors and Genetic Alliance representatives to discuss ways to continue to work together.

Expanding Our Reach

Liaisons represented NSGC at several meetings this fall: **Maureen Smith** was NSGC's representative at a meeting in Cold Spring Harbor NY to address the future training of medical geneticists. **Nancy Warren** attended the Association of Schools of Allied Health Professions and reported on the significance of online education and growing awareness of genetic counseling as a profession. **Jennifer Farmer** spearheaded efforts to display our booth at the American Association of Family Physicians annual meeting.

Looking Ahead

The year ahead promises to be busy. Your Board is working hard to formalize plans for incorporating an NSGC Foundation. We will continue our forward momentum on billing and reimbursement. Several states will receive funding to assist in licensure efforts. **Nancy Callanan** and our regional representatives will begin a leadership initiative, and we will make efforts to increase our visibility to undergraduate students. I hope we can begin to address the monumental issue of health disparities as they relate to genetic counseling, including minority recruitment and retention.

I look forward to representing you in 2005. To discuss issues or get involved, please feel free to contact me. Happy holidays, and best wishes for a safe and happy new year! ❖

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A Case of Enhanced Liability Risk: An Unexpected Legal Outcome

Sandra B. Factor, JD, MS

All medical professionals are aware of medical malpractice. The following case involving an unanticipated medical event with a negative legal outcome shows how medical liability could extend to genetic counselors working in prenatal and pediatric care.

Facts of the Case

On May 22, 2004, the Minnesota Supreme Court ruled to bring to trial three doctors who failed to diagnose a child with fragile X and warn the mother of the potential for having another affected child. The Court determined that physicians have a duty beyond the patient regarding genetic test results and that a genetic risk extends to parents who "may be harmed by a breach of that duty."

The case was brought by the mother and her current husband when their son was born with fragile X – ten years after the birth of her daughter, from a previous marriage, who had developmental delay. When the son was born, the mother was tested for fragile X and found to be a carrier. The woman and her husband claimed they would not have conceived their son if they had known these results before.

Missed Diagnosis

When the daughter was diagnosed with developmental delay at age three, their pediatrician said he would test for fragile X and other possible causes. Fragile X was not included in the tests, however, and the pediatrician reported that all results were normal.

Later, two neurologists treated the daughter for developmental delay but did not suggest further genetic testing. They also were named as defendants. If they had relied on the results of the previous genetic tests, their liability for failure to diagnose may be more defensible, but they are not immunized from liability by relying on another doctor's opinion.

The Ruling

The defendants claimed the case should be dismissed on two grounds:

- the statute of limitations had run out, i.e. the lawsuit was too old, and
- the daughter was the patient and thus the mother and husband did not have "standing" to act as plaintiffs and sue on their own behalves.

While it is true that the daughter was not the plaintiff and the alleged failure to diagnose did not cause her fragile X, the mother's argument seemed to support a failure by the doctors to diagnose the daughter's condition and prevent the "wrongful life" of the son.

In bringing the doctors to trial, the Court is setting a precedent allowing parents of genetically affected children to sue for harm done to them in bearing another affected child. The ruling also extends the time frame for litigation, allowing relatives and descendants to claim standing years after a medical/genetic evaluation. The Court has, through "judge-made law," carved out an exception for genetic conditions that is not currently enacted by the Minnesota state legislature.

Definitions

Medical malpractice is an act of negligence where the plaintiff claims that harm was caused by the defendant and demands monetary compensation. In medical malpractice cases, the plaintiff must prove four elements:

1) duty of care was owed by the physician to the patient;
2) the physician violated the applicable standard of care;
3) the patient suffered a compensable injury; and
4) such injury was caused by the substandard conduct.

Traditionally, this injury means physical harm to the patient, but recently courts have been recognizing cases of emotional harm.

Protecting Yourself

Medical professionals know that bad outcomes may occur without negligent care. These risks should be thoroughly discussed prior to treatment, and patients should provide informed consent to proceed despite acknowledging in writing the risks.

Because obstetricians, perinatologists, neonatologists, genetic counselors and pediatricians are at high risk for liability exposure, maximum liability insurance is advised. Legal "infancy" varies by state but may extend to 21 years of age, meaning medical professionals may be sued years into the future.

Conclusion

Over time, case law evolves. Plaintiffs win cases that previously had not been considered. Medical practices change with new therapies and diagnostic tools. For genetic counselors and all medical professionals, the highest standards of care must be maintained for their own benefit as well as for their patients. ❖

☞ **Sandy Factor,** Chair, Legal SIG; engravitas@aol.com

Career Watch

Prenatal Pediatrics Expands the GC's Role

Karlla W. Brigatti, MS

Prenatal diagnosis once was limited to amniocentesis or CVS. Yet recent advances in proteomics, genomics and stem cell research have placed *in utero* treatment of some genetic conditions on the foreseeable horizon. Improved technologies in ultrasound and the use of ultrafast MRI have increased our ability to visualize fetal anatomy, accurately diagnose structural anomalies and better predict prognoses.

Karlla W. Brigatti, MS

Throughout the US, a handful of tertiary care centers specialize in this cutting-edge approach to prenatal diagnosis, pregnancy management, planned delivery and specialized neonatal care. One such program is the Center for Prenatal Pediatrics at Columbia University Medical Center in New York, which was founded earlier this year in part with a grant from the March of Dimes. I function as the Clinical Care Coordinator, and the genetic counselor.

Prenatal Pediatrics

The term "prenatal pediatrics" illustrates the principle that the care of babies with abnormalities begins during the prenatal period. This integrated approach requires the involvement of both prenatal and postnatal specialists such as neonatologists, pediatric cardiologists, pediatric surgeons, geneticists and many others.

At the Center, our philosophy is the integration of prenatal and neonatal care, education and research. Care begins when the anomaly or syndrome is first identified. Our team works together to counsel the patient and give her and her family a perspective on prognosis, diagnosis and treatment. Each patient receives a tailored program of prenatal testing and specialist consultations, all conveniently scheduled in one day. Once the patient has been evaluated, we host a comprehensive care conference with the patient and family to summarize the day's results. We also devise a follow-up prenatal and neonatal program, in coordination with the referring doctors.

More than Just a Coordinator

My training as a genetic counselor is indispensable in my job as Clinical Care Coordinator. My experiences in both prenatal and pediatric settings have provided an ideal skill set. The Center's emphasis on psychosocial counseling and patient advocacy has turned a coordinator position into something much more meaningful and useful to patients. My understanding of the complexity of the medical information and how to communicate these ideas to patients in a personalized context has been a large part of our success.

Science and Psychology

People sometimes ask if my job gets to me, since all Center patients are expecting babies that will need significant follow-up, many with incredibly poor odds. While my sense of pregnancy revolves around cases of congenital diaphragmatic hernia, hypoplastic left heart, twin-to-twin transfusion and hydrocephalus, I am amazed and impressed by the strength and grace parents exhibit in these situations. When babies go home, I celebrate with the parents and medical team. When babies don't get that chance, we all feel the loss. I take comfort that, no matter the outcome, parents believe that the Center makes a difference for them. I also appreciate the marriage of science and psychology that the Center achieves on a daily basis. ❖

Free On-line Seminars on Lysosomal Storage Disorders

Exceptional Parent magazine is hosting two free CME/CEU-accredited on-line teleconferences titled, "Pediatric Disabilities in the Lysosomal Storage Disorders, Pompe Disease and MPS I," on January 14 and 28 from 7-9 pm EST. Presenters are **Drs. Priya Kishnani, Debra Day-Salvatore** and **Joseph Muenzer**. **Dr. Alan Percy** moderates.

Session one will overview Pompe disease and MPS I, including clinical manifestations, diagnostics and treatments. Session two will review emerging therapies, care and treatment. The seminars are endorsed by the Child Neurology Foundation and the National MPS Society. Participants must pre-register. ❖

- ☞ www.eparent.com; 800-372-7368, ext. 203
- ☞ **Bridget M. Lyne,** 201-489-4111, ext. 211; blyne@eparent.com

MILITARY, *from page 1*

about two million family members, or "dependents" of active duty service members. These figures do not include veterans, military retirees or their families. Thus, it is very likely that, at some point, a genetic counselor will interact with a military family.

RECOGNIZING RANK

One of the most obvious differences between civilian and military populations is the role of rank. Military rank is an integral system of seniority and command. Rank is determined by years of service, education and past performance evaluations. Housing options, pay rates and job opportunities are partially determined by rank.

When addressing military personnel, use of an individual's rank is a sign of respect. It often is possible to determine an individual's rank by his or her uniform. It also is appropriate to ask how an individual prefers to be addressed, as some personnel prefer less formality when interacting with civilian health care providers.

PERMANENT CHANGE OF STATION

Frequent relocations, known as a permanent change of station (PCS), are an accepted part of military life that present challenges for genetic counselors. A military family's support system is different and often more limited than a civilian's. Relatives and long time friends are seldom nearby. Often, the military becomes a surrogate family, with higher-ranking individuals serving as advisors and mentors for younger troops.

In addition, frequent PCSs can interrupt continuity of medical care. Medical records may be difficult to track down. Also, with every move, a family's medical needs must be met by a new team of providers. For families of children with special needs, such changes can be complicated and frustrating. It is important to assess the family's support system and to offer resources such as support groups and family contacts whenever possible.

DEPLOYMENT

Another issue faced by many military families, particularly in recent years, is deployment to a foreign country or a war zone. From a practical standpoint, family history of the deployed individual will be more difficult for a genetic counselor to obtain. Also, the deployed individual is not available for genetic testing, a particularly problematic situation when considering prenatal carrier screening.

From a psychosocial standpoint, the family of a deployed service member may be under a great deal of stress. Everyday tasks may become more burdensome. Communication between relatives may be sporadic and unpredictable. If the individual is deployed to a war zone, the family also may be worried for their loved one's safety. Under such strains, receiving a new genetic diagnosis or facing a critical decision regarding a pregnancy can be devastating. Assessing the family's support and coping mechanisms and providing appropriate resources are critical.

RESPECT AND REFLECTION

Upon hearing that a person in the military is being deployed, most civilians would respond by expressing sorrow. After all, deployment implies a geographic separation of the family and may involve transfer to a dangerous location. However, such empathetic statements may not be helpful or welcomed. Many service members and their families accept deployment in a matter-of-fact manner. The service member may view deployment as an opportunity to serve his or her country, the very reason he or she joined the military. Others may view deployment just as a part of military life or may feel it inappropriate to express their emotional response within a medical setting. In general, it is most useful simply to reflect the family's response to the deployment.

PERSONAL REFLECTIONS

My husband is a Captain in the US Air Force. The greatest challenge in being a military wife has been our frequent moves and their impact on my career in genetic counseling. I have learned to create job-related opportunities, as I recognize that my time in any one city is limited. Participating in the NSGC also has been critical in keeping me up-to-date within my field as well as connected with my genetic counseling colleagues. My NSGC involvement has been the only constant throughout my career.

Despite the challenges it presents to my professional career, the military has provided us with many unique experiences, including opportunities to travel within the US and overseas and to meet many new and interesting friends and genetic counseling clients. I am impressed by the work that my husband and his colleagues do, as well as the sacrifices made by our men and women in uniform. ❖

This article is the first in a new series of Voices that opens a window on the diversity of our profession. Listen to members of our Society from different backgrounds and cultures and at different stages of life describe what it is to be a genetic counselor.

The Voice of a New Grad: Thoughts on Our Profession

Jessie Ray, MS

My second year in graduate training was a countdown to graduation. I had no misconceptions of a six-figure salary; I was aware of the steep road ahead. Yet I couldn't wait to dive right in to my profession of choice. Now that I have taken that plunge and am working in the field, with its seemingly endless potential yet its struggle to cement itself in the medical forefront, my experiences have fostered a new perspective.

The Luxury of Time

As a student, having time to analyze the impact genetic information can have on patients was a luxury. I had no idea of the time commitment required to fully prepare for, counsel and follow-up on each patient. I now realize that the number of patients I can effectively see each week is much less than expected.

Amidst the time constraints, I strive to create a bond with each patient. I have a new appreciation for family systems and have witnessed the far reaching impact of genetic information. Helping patients work through their decisions, whether they are pursuing testing or risk-reduction options, has been extremely rewarding.

Colleague Support

I am the only genetic counselor in the cancer center where I work. I have expanded my circle of colleagues from the genetic counselors of graduate school to the physicians, nurses, social workers, therapists and nutritionists I interact with daily. I have become part of an integrated team, where every member has a role in the well-rounded care of the patient. While I am grateful for this support, being the only counselor at times puts an overwhelming responsibility on my shoulders.

Continual Lessons

While I appreciate my graduate training, I continue to learn with each patient, each interaction with a referring physician, each struggle with an insurance company and each genetic test result. I believe I have signed on to a profession that will require continual effort. I also expect, however, that it will foster a lifetime of learning and diversity as well as satisfaction and possibility. ❖

☞ To contact *Voices*...
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Student Corner

Calling All Students: Getting Involved with NSGC

Stephanie Herbert, BA

As the first half of the school year comes to an end, genetic counseling students are getting the chance to take a breath. Between placements, classes and thesis projects, there is often little time for more commitments, such as becoming involved with NSGC.

Despite busy schedules, however, students still want to share with professionals in the field. That is the goal of this new column: to provide students an opportunity to get involved. Did you land a unique internship, run a grant-funded project or complete a novel thesis? Did you overcome a counseling or supervisory challenge? Fellow students and practicing counselors alike can learn from these experiences.

Highlights

This year's AEC in Washington DC was undoubtedly a highlight of most genetic counseling students' fall semesters. **Penelope Roberts,** from Brandeis University, said, "It was great to see the vast size of our organization and the number of interesting talks that were available. I had no idea how big the NSGC was until I walked into the ballroom and saw the enormous number of participants."

Networking

Brianne Williams, from Northwestern University, describes her experience as "a whirlwind of busyness... between catching up with supervisors and students from other programs, to attending the presentations, sneaking off to the Smithsonian, interviewing and networking. I slept less than I do during crunch-time at the end of a semester. But the running around was worth it because I came back knowing more friendly faces in NSGC and with some job prospects…." ❖

☞ sherbert@brandeis.edu

AEC 2005 – Los Angeles Here We Come!

When: November 12 – 15, 2005

Where: The Westin Century Plaza Hotel & Spa
www.westincenturyplaza.com

Though it seems we just returned from the recent NSGC meeting in Washington, planning is underway for the 2005 Annual Education Conference (AEC) in Los Angeles CA. Our goal is to provide 24 hours (2.4 CEU) of educational sessions with extra time for committee and SIG meetings, reunions and breathing room! Additional CEUs will be available with sponsored breakfasts/lunches.

The AEC will begin on Saturday afternoon and adjourn on Tuesday. Sunday, Monday and Tuesday will be full days. Considerations for this schedule included comments from the 2004 AEC evaluations, east coast flight times and LA rush hour traffic.

NSGC members will receive the conference brochure in early spring. Information also will be posted on the NSGC website and provided to other professional organizations, such as the International Society of Nurses in Genetics and the Canadian Association of Genetic Counselors. Stay tuned for information about organized tours of the city and other activities!

Short Course

There will be one short course offered on the Thursday and Friday before the AEC titled, "Reaching Our Clients: Risk Perception and Communication in Genetic Counseling Practice." All areas of genetic counseling will be addressed.

Abstract Deadlines

Abstracts of interest to genetic counseors will be accepted for consideration as platform or poster presentations between April 4 and June 3, 2005. Students, non-members and full members are encouraged to apply. Monetary awards will be presented for best full and student member abstracts. For submission instructions, visit www.nsgc.org.

Your Voice Counts

It's not too late to volunteer! Contact any of the conference chairs to help make the conference a success.

2005 AEC Planning Committee

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2004 AEC Award Winners

NSGC would like to recognize the 23rd Annual Education Conference registrants who won give-aways at this year's meeting in Washington DC.

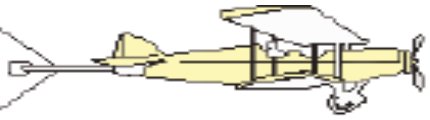
For the second year, NSGC has acknowledged members with Amazon.com gift certificates in our "Leading Voices" program. Throughout the year, the Board of Directors identifies members who have taken on leadership roles and valued projects for NSGC and sends these names to the Executive Office. Immediately prior to our Open Mike session, 10 names are drawn, and each receives a $20 Amazon.com gift certificate. This year's winners were: **Judyth Greenbaum, Nancy Kramer, Lisa Dellefave, Norma Chow, Michelle Fox, Ed Kloza, Margo Grady, Kristin Niendorf, Jennie Fieger** and **Karen Wolff.**

NSGC's "Publisher's Special" invites publishers to submit books and items to be displayed at our conference and given away in a drawing. This year the donors were the American Psychological Association, Cold Spring Harbor Press and chromi.com. The winners of this drawing were: **Maggie Dewhurst, Michelle Martin, Jacqueline Ogilvie, Sarah Lewis, Cindy Soliday, Erin Carter, Kate Kramer, Betsy Gettig** and **Bonnie Liebers.**

In a final drawing, NSGC offered a complete set of the *Journal of Genetic Counseling*, volumes 1 – 12, in hard copy. The winner was **Beverly Yashar.**

Congratulations to all 2004 winners, and thanks to everyone who donated their time, energy and efforts to NSGC! ❖

MEDIA WATCH



Angela Geist, MS and Roxanne Ruzicka, MS

June 5 – *British Medical Journal*, "New Law Forces Italian Couple with Genetic Disease to Implant all their IVF Embryos"

An Italian judge ordered a sterile couple to transfer all of their embryos obtained with assisted reproduction techniques (ART) even though both parents carry the recessive gene for beta thalassemia. The law, approved earlier this year, bans freezing and destroying embryos, limits the number of oocytes fertilized, requires all created embryos to be transferred and restricts the use of ART to sterile couples. Hundreds of non-sterile couples, fearful of having babies with genetic illnesses, have started to go abroad, as have many specialists and researchers.

August 27 – *Jewish News Weekly of Northern California*, "Heartbreak of Genetic Disease"

An article discussed the importance of screening for genetic disorders in the Jewish population. California funds prenatal and newborn screening programs, yet these do not screen for conditions prevalent among Jews. Therefore, "pre-pregnancy genetic counseling is the answer." The success of gene therapy in treating Gaucher disease and community education in reducing the incidence of Tay-Sachs disease were mentioned.

August 31 – *New York Times*, "The Havoc of an Undetected Extra Chromosome"

This story discussed the symptoms and management of 47,XXY. One mother of an affected child mentioned that her doctor was not familiar with the condition. There was no mention of genetic counseling.

Fall '04 – *Cure* magazine, "Screening for Colorectal Cancer"

A spotlight on colon cancer addressed colon cancer screening to enable early detection and treatment. It reviewed the different types of screening and mentioned some hereditary colorectal cancer syndromes. The article contained a link for the NSGC website.

September 6 – *National Law Journal*, "Genetic Testing Maps New Legal Turf; Doctors' Liability Grows as Tests are More Widely Used"

This article described how genetic testing has expanded physicians' liability regarding counseling patients about hereditary disorders. The article reviewed recent court decisions that tried to clarify physicians' duty to relay information from genetic testing. Cases ranged from risks to other family members to whether parents can collect damages for having a child with genetic defects due to a physician's failure to identify the likelihood of the disorder.

September 29 – *The Seattle Times*, "Genetics, History Spelled Cancer, so She Opted for Preventive Mastectomy"

This comprehensive article followed a woman through her evaluation for breast cancer risk, her fear of cancer, her decision to have BRCA testing and her subsequent prophylactic mastectomies. The article mentioned **Lisa Amacker North** and reviewed the genetic counseling and testing process.

October 4 – *Playboy* magazine, "Who's Your Daddy?"

This item stated that,"basing their findings on the results of tests for inheritable diseases, genetic counselors estimate that 10% of children in America, unbeknownst to their presumed fathers, were actually sired by another man."

October 28 – Discovery Health Channel, "Breast Cancer Legacy"

This documentary illustrated the medical, psychological, ethical and interpersonal issues faced by women confronting cancer predisposition genetic testing. **Shelly Cummings** was featured. **Laura Dudlicek** and other medical professionals attended panel discussions held in several cities before the show was aired.

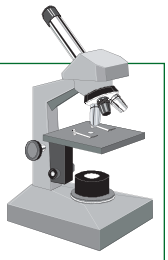
November 2 – *Los Angeles Times*, "Sisters Linked in the Lab"

This story described how families are using DNA testing to establish biological links between adopted children. While ethicists worry that the results can be unsettling, two families shared the joy they experienced when their daughters discovered they were half-sisters.

November 8 – *American Medical News*, "Genetic Testing Issues Land in Primary Care Practices"

A family physician was featured on how genetic testing and difficult patient decisions are increasing within primary care practices. When asked by a patient with a family history of breast cancer if she should have BRCA testing, he replied, "Mostly, what I've been telling her is I don't know." This doctor attended sessions on genetic testing at a recent American Academy of Family Physicians conference. ❖

Research Network



✓ NIH study on Hearing Loss and EVA

The Department of Health and Human Services, National Institutes of Health (NIH), National Institute on Deafness and Other Communication Disorders, is seeking individuals of all ages with enlargement of the vestibular aqueduct (EVA) to participate in a research study. The study's goal is to better understand the causes of EVA. Individuals may qualify if they have hearing loss and EVA in one or both ears. Individuals may not qualify if they have previously been diagnosed with BOR syndrome. We also are interested in enrolling siblings and parents of individuals with EVA.

Participation requires travel to the NIH Clinical Center in Bethesda MD for outpatient evaluations that require approximately six hours per day for three days. There is no cost to participate or to receive any of the study associated evaluations. Travel, lodging and food can be subsidized. A blood sample is requested for genetic testing. Participants will receive the results of study associated evaluations.

This study is NIH protocol #01-DC-0228, "Clinical and Molecular Analysis of Hearing Impairment Associated with Enlarged Vestibular Aqueducts." This study is carried out in compliance with testing and safety standards of the US Department of Health and Human Services.

☞ **Anne Madeo, MS,** 301-435-1574; madeoa@nidcd.nih.gov

✓ Noonan Syndrome and Related Disorders

The Center for Genetics and Genomics at the Harvard Medical School and Partners Healthcare System is conducting a new research study for patients with Noonan syndrome, isolated pulmonary valve disease or isolated hypertrophic cardiomyopathy. The goal of this study is to understand the gene changes that can lead to Noonan syndrome and related disorders. Enrollment involves a physical exam, medical history and family history. A blood sample is required. Patients who have never had a cardiac ultrasound or renal ultrasound will have these studies completed. With patient consent, we will establish cell cultures and DNA to be stored. The results of all testing will be made available to the patient and referring physician.

☞ **Taryn Schiripo, MS,** 617-525-4490; tschiripo@partners.org
☞ **Amy Roberts, MD,** 617-525-4485; aeroberts@partners.org

✓ Telomere Study at Emory University

The Telomere Research Project, led by **Drs. David Ledbetter** and **Christa Lese Martin** in the Department of Human Genetics at Emory University, is recruiting individuals and families with an identified telomere abnormality (that is not cytogenetically visible) for fine mapping studies to establish genotype/phenotype correlations. Two categories of telomere rearrangements are of interest: 1) those causative of an individual's phenotype and 2) those identified in an affected proband but subsequently found to be carried by a phenotypically normal parent (benign variants). ❖

☞ **Sara Cooper, MS,** (404) 727-7098; scooper@genetics.emory.edu

For Your Practice

Genetic and Rare Diseases Center

Janine Lewis, MS

The National Institutes of Health (NIH) offers health professionals and the public free assistance in finding current and accurate information about genetic and rare diseases through the Genetic and Rare Diseases (GARD) Information Center.

Keeping pace with the growing body of scientific knowledge about genetic and rare diseases is an uphill battle for many health care professionals. Another major challenge is finding the time and resources to clearly convey this information to patients.

For three years, GARD has provided a bridge between information seekers and reliable resources for genetic and rare diseases. Experienced information specialists are available to answer questions between 12 pm and 6 pm EST, Monday through Friday. Written responses usually are provided within five business days.

For genetic counselors and health care providers, GARD can help prepare for an upcoming case or reinforce or expand upon information already provided to patients. To date, GARD has responded to more than 9,000 inquiries. GARD does not provide genetic counseling or diagnoses but refers inquirers to local genetic services when appropriate. ❖

☞ GARD, 888-205-2311; TTY: 888-205-3223; GARDinfo@nih.gov; www.genome.gov/Health/GARD

☞ ORD (Office for Rare Disorders); rarediseases.info.nih.gov/html/resources/info_cntr.html

EXPECTING ADAM

By: **Martha Beck**
Publisher: Berkley Books, New York, 328 pp., $13.95 (paperback)
Reviewed by: **Autumn Tansky, BS**

Expecting Adam is an inspirational, self-authored narrative tracing the dramatic change in Martha and John Beck's lives from the conception to the birth of their son, Adam, who has Down syndrome.

Discovering that a pregnancy is affected with Down syndrome can profoundly impact any couple's life. This diagnosis is especially poignant for the Becks – type A personalities dividing their time between taking classes and teaching at Harvard, consulting in Singapore and caring for their 18-month-old daughter. Each day is controlled by a strict schedule with the goal of obtaining their Harvard PhDs and achieving their fullest professional potential.

Even before prenatal testing, Martha and John know this pregnancy is unique. Martha writes of the extraordinary and supernatural events she and John experience. A recurrent spiritual presence, invisible helpers and an ability to mentally join John in Singapore while physically being in Boston are integral parts of her perseverance while expecting Adam.

Martha is honest regarding the negative response and, at times, overt resistance to their decision to continue the pregnancy, particularly by many of their teachers and colleagues and the University Health Service's prenatal doctors. Although at times challenging to believe, this book encourages the reader to question the validity of societal ideals and rediscover that life's greatest lessons are not taught in a classroom. I would recommend this book to genetic counselors, students, parents of children with Down syndrome and those willing to redefine the concepts of disability and normality. ❖

FAMILIAL BREAST AND OVARIAN CANCER: GENETICS, SCREENING AND MANAGEMENT

Edited by: **Patrick J. Morrison, Shirley V. Hodgson** and **Neva E. Haites**
Publisher: Cambridge University Press, Cambridge UK, 400 pp., $95 (hardback)
Reviewed by: **Susan Armel, MS**

The difficulty with writing a cancer genetics book is that by the time it is published it often is outdated. Despite this conflict, *Familial Breast and Ovarian Cancer* provides a good summary of cancer genetics, screening and management for BRCA1 and BRCA2 mutation carriers.

The text is organized into three parts: 1) molecular biology and natural history, 2) screening and 3) management. Each section is divided into chapters that are written by different authors.

NATURAL HISTORY

Part 1: The book begins with a review of the molecular biology and natural history of hereditary breast and ovarian cancer. Though some statistics are outdated, it includes a nice review of Cowden syndrome and addresses many important issues encountered when counseling high-risk families. The first section concludes with a review of risk assessment models and lists appropriate Internet sites.

THE GENETIC COUNSELOR'S ROLE

Part 2: In this section, the book features cancer genetic services in the UK. The education of primary care physicians, the evaluation of services and referral guidelines are compared in areas throughout Europe. The genetic counselor's role is briefly discussed, primarily in the context of helping in pedigree construction and evaluation. The text differentiates that physicians should counsel individuals eligible for genetic testing, and genetic counselors should counsel low- or medium-risk individuals. While perhaps frustrating to read, at least it is encouraging that our profession is recognized by the authors.

STRATEGIES AND UPTAKE

Part 3: The book concludes with a section on risk reduction strategies and screening options available to high-risk women. The authors discuss the psychosocial aspects of genetic counseling for breast and ovarian cancer, the ethical, social and insurance issues, and the uptake of BRCA1 and BRCA2 genetic testing and measurement tools in the UK. The authors hypothesize about the future of cancer genetic testing and counseling and the possible impact of gene therapy. This book is applicable to a wide audience due to the ease in reading and many illustrative tables. While written from the European perspective, most of it is applicable regardless of location. This resource should be a useful addition to any cancer genetics library. ❖

Special Interest Groups in 2005

SIGs are a way for members who share professional interests to network with each other. Here is a list of active SIGs for 2005. Please contact SIG leaders for information on current projects and how to get involved!

ART/Infertility	**Kari Danziger;** danziger@urol.ucsf.edu
Cardiovascular Genetics	**Deborah McDermott;** dam2001@med.cornell.edu **Amy Sturm;** sturm-1@medctr.osu.edu
Clinical Supervisors	**Sue Demsey;** sue.a.demsey@kp.org
Familial Cancer Risk Counselors	**Cecelia Bellcross;** cecelia.bellcross@deancare.com **Scott Weissman*;** sweissman@eng.org
Industry	**Cynthia Frye*;** cfrye@myriad.com; **Kathleen O'Connor*;** koconnor@reprogen.com
Legal	**Sandra Factor;** engravitas@aol.com
Metabolism/Lysosomal Storage Disorders	**Juliann Stevens-Harvey;** JMSHarvey@aol.com
Neurogenetics	**Aideen McInerney-Leo*;** amcinern@nhgri.nih.gov **Malia Rumbaugh*;** maliarum@u.washington.edu
Pediatrics	TBA
Prenatal	**Sarah Noblin;** Sarah.J.Noblin@uth.tmc.edu
Private Practice	**Kelly Donahue*;** kdonahue@wpahs.org
Psychiatric Disorders	**Holly Peay;** hlpeay@nchpeg.org
Public Health	**Suellen Hopfer*;** sxh343@psu.edu
Research	TBA
Telegenetics	**Angela Arnold;** angela.arnold@royalfree.nhs.uk **Becky Butler;** bbutler@uams.edu

**Indicates new chairperson*

NSGC Announces Licensure Grant Winners

Earlier this year, NSGC established a grant for members working to pass state licensure for genetic counselors. In 2004, $3,000 was committed. Approximately $15,000 is available in 2005, to be awarded in two cycles.

How and When to Apply

Applicants are asked to outline their plan for organizing genetic counselors' work toward licensure in their state, review their state's history of licensure efforts, document the need for grant support and describe how award money will be used. Only one application is accepted per state. Application instructions were emailed to all NSGC members and are posted on the NSGC website. Click on "Licensure" from the members area. Notice of the second cycle of applications for 2005 awards will be posted in February.

And the Winners Are...

2004 Award Winners:

- Florida (**Dan Riconda)**
- Illinois (**Judy Miller**)
- Texas (**Aimee Tucker Williams**)

2005 1st cycle Award Winners:

- Massachusetts (**Kristen Shannon**)
- Tennessee (**Amy Crunk**)

Licensure Committee

The 2004/2005 Licensure Grants Committee includes: **Rob Pilarski,** Chair; **Karin Dent, Sara Goldman, Cheryl Harper, Carrie Haverty, Chris Miller** and **Jennifer Farmer** (ex officio).❖

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Billing & Reimbursement Primer Now Publicly Available

In response to increasing public inquiries regarding billing and reimbursement (B&R) for genetic counseling services, NSGC's Billing & Reimbursement Primer is now available on the public portion of our website.

Useful Resource

The primer has become increasingly useful to employers and genetic counselors as they work together to determine the best methods for institutional billing for genetic services. Developed by members of the Genetic Services Committee and the Billing and Reimbursement Subcommittee, the primer contains information and resources available for understanding the current state of billing for genetic services.

Increasing Recognition

Making the primer publicly available will hopefully increase institutional and payer recognition of B&R-related issues impacting genetic services. Access the primer at www.nsgc.org and click the link entitled "Resources for Other Professionals."

If you have utilized the B&R Primer to successfully establish billing for genetic services at your institution, the B&R Subcommittee would like to hear from you. Your story may help others negotiate billing in their practice.

☞ **Aimee Tucker Williams,**
B&R Subcommitee Chair;
aimee.j.tucker@uth.tmc.edu.

NSGC Website Watch
For the Members, By the Members

Third in a Four Part Series
Debbie Keelean-Fuller, MS

The members' section of the webpage is dedicated to YOU – whether it be information on the Board or a SIG, viewing the latest version of *PGC* or *JGC*, downloading a PowerPoint presentation for a talk you'll be giving or reviewing tools for your clinical practice. This article will highlight some exciting changes to the NSGC website and will provide you with guidelines on how to post new information on the web.

Additions to the Website

Do you know where to find information about your region or past professional status surveys? Do you recall an interesting topic from the members' site but can't remember which SIG or committee posted it? Use the search function! You can now search the entire NSGC website. The search tool is found in the upper right-hand corner of every page on both the public and members' side. From the members' side, the search can be refined after the initial inquiry. And don't forget, if searching for information, *PGC* is now indexed back to the original issue in 1979. Just click on the *PGC* link on the NSGC home page, under "Members News."

Posting New Info on the Web

Here are some general guidelines when you have new information to post on the web. Text content for web pages generally can be written in standard word processing, such as Microsoft Word. Content should be forwarded to me, your new web editor, along with a list of instructions concerning placement of the new information and any links that are needed within the document. We can help you with questions regarding formatting and posting. Any substantial additions to the website need to be reviewed by the NSGC Publications Subcommittee for editing and consistency with NSGC standards. Timing for this process varies depending on editing needs, project size and queue of pending projects.

New Editor at Your Service

We strive to provide you with the best website that fits your needs. However, improvement of a website is an ongoing process. If you would like to help with the website, or if you have constructive suggestions for what you would like to see on the web, please contact me. ❖

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Next month: *Spotlighting Counselors: Providing a Public Face for our Website*