nsgc

PERSPECTIVES IN GENETIC COUNSELING

newsletter of the National Society of Genetic Counselors, Inc.

Vol 14, No. 1 Spring 1992

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- Jane Engelberg Memorial Fellowship Applications — April 1
- Call for Nominations — April 3
- NSGC Membership Dues Last Call for FY92 — April 20
- Special Projects Fund Applications — May 15
- Abstracts for Posters and Presented Papers, Annual Educational Conference, San Francisco — May 29

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DIAGNOSTIC TESTING FOR FRAGILE X SYNDROME

by Amy Cronister Silverman, M.S., Vivigen, Inc., Santa Fe, NM

For more than a decade, the fragile X syndrome has confounded geneticists. Baffling recurrence risks, variable expressivity, non-penetrance and imperfect carrier and prenatal diagnostic testing have toyed with what we knew about Mendelian inheritance. Within the past year, rapid advances, including localization of the fragile X (FMR-1) gene have occurred in our understanding of the fragile X region at the molecular level. The FMR-1 gene has been shown to contain a variable repetitive sequence of DNA. Using Southern blot analysis or PCR, insertion lengths and methylation patterns can be detected and analyzed to distinguish heterozygous fragile X females and transmitting males from normal individuals, and those affected with fragile X syndrome can be distinguished from individuals with other forms of mental retardation.

DNA mutation analysis is more sensitive than cytogenetics when testing at-risk intellectually normal relatives of known fragile X individuals. Prenatal fragile X cytogenetic analysis alone can detect approximately 91-95% of affected pregnancies. Complementary prenatal DNA linkage studies may increase reliability to 99%, but inconclusive results have caused problems. Direct DNA mutation analysis can significantly enhance the accuracy of prenatal diagnostic testing because it can detect females with the premutation (who would be at low risk for mental impairment), non-

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GUIDING PRINCIPLES, RESOLUTIONS CLARIFY STANCE

by Shane Palmer, M.S., Dept Environment, Health & Natural Resources, Washington, NC

The Social Issues Committee has the responsibility for making social policy recommendations to the NSGC Board of Directors and membership. One of its first activities was to present the Reproductive Freedom resolution for membership vote, which passed in 1987. Next, the Board appointed a legislative liaison, and an ongoing education forum with a legislative column in *Perspectives* was created. The resolution has been a valuable tool for our Social Issues Committee and the legislative liaison. Subsequent to that, this committee constructed additional Guiding Principles and Resolutions. These were initially presented to the Board of Directors in July 1990.

It is standard practice for medical professional societies to have policies which provide guidance and definition. As an example, the American College of Obstetricians and Gynecologists has adopted policy statements about fetal tissue research, access to abortion and MSAFP screening. The American Academy of Pediatrics has adopted policy statements about nutrition, circumcision and, most recently, support for a ban on hand guns and assault

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PRIVATE PRACTICE GENETIC COUNSELING

Independence. Flexibility. Diversity. Family. Office administration. Billing control. These words can be used to describe the life of genetic counselors who have carved out an exciting alternative career path in genetic counseling.

Medicine is an evolutionary science, and genetics seems to be on the forefront of the change. Some of the following changes have helped open the door for the private practice genetic counselor.

- **EXPLOSION OF GENETIC INFORMATION:** Demand for genetic counseling will grow with each new discovery.
- **EMERGENCE OF THE PARAPROFESSIONAL:** As demands on physicians grew, so did the acceptance of the paraprofessional such as the midwife and nurse practitioner.
- **MEDICAL MARKETING:** Marketing is now an essential part of hospital and private offices, and comprehensive services make an attractive part of a marketing program.
- **SHORTAGE/MALDISTRIBUTION OF TRAINED GENETIC COUNSELORS:** Even in established genetic centers, there are often not enough trained individuals to fill positions. Less traditional situations that could benefit from a genetic counselor have been doing without.

Each of us has recognized these changes and carved out unique niches in our communities. Even with diverse experience and geographical backgrounds, we agree that there are pluses and minuses.

POSITIVE ASPECTS

Personal Benefits: The various activities of private practice include structuring a personal schedule and managing projects from planning to completion. The mix of patient counseling, educational projects, marketing, office administration and free time is dependent on the individual's own needs and interests. The individual with ability and perseverance can easily generate more income per hour than most traditional positions. The operative word is *per hour*, since personal control of a schedule allows the number of hours to vary. This variable schedule can also be tailored to suit personal and family needs.

Mobility: The main beneficiaries of our services are the community hospitals or private physician offices already performing prenatal diagnostic techniques. These smaller facilities cannot afford a full time genetic counselor. However, a counselor willing to work at several locations on an "as needed" basis can provide several benefits:

- the patient feels comfortable in a familiar setting;
- the available in house services have been expanded,
- staff members learn the value of genetic counseling and interact directly with the counselor.

This independence can also help the genetic centers. Qualified genetic counselors willing to work part time or flexible hours can assist with the burdens of staff vacations, maternity leave and conferences.

Expanding Professional Options: The potential to "freelance" enables genetic counselors to follow career interests on their own schedule rather than leave the profession because of limited career advancement possibilities or a desire to raise a family.

> ***"The potential to 'freelance' enables genetic counselors to follow career interests on their own schedule..."***

NEGATIVE ASPECTS

Lack of security and camaraderie: There is comfort in being surrounded by other genetic counselors, medical geneticists and expensive reference materials. Private practice counselors need to establish relationships with professionals who will review cases and be a network for discussion. Medical literature must be reviewed regularly and with a greater intensity to compensate for the lack of readily available knowledge from staff meetings and conferences. Training programs do not teach many of the skills, such as marketing, office management and accounting, all essential to private practice.

Income: Before the ability to regulate your income, there is a period of no income. Set up costs such as malpractice insurance, business cards, license and incorporation fees consume reserves before an income is realized.

Benefits: Some of the benefits counselors have been striving for, conference reimbursement and travel expenses, are not part of the private practice scenario. In addition, income can be irregular and personal health insurance can be costly.

Negative reactions from colleagues: We have all encountered negative reactions from colleagues in traditional settings. One concern is that we are taking patients away from their centers, when in actuality, many of our patients are from populations not previously served. There is also the insinuation that we are not as well informed since we are not in an academic center, or that we are seeing inappropriate cases (ones that should be seen by a medical geneticists.) We are all aware of our limits and, in fact, often act as a referral base for traditional genetic centers. Our emphasis is on service and maintaining the standard of care.

Overall, private practice genetic counseling can meet patient, professional and personal needs and is an important alternative to the traditional genetic counselor positions available.

by Debra Han, M.P.H., Susan Mundt, M.P.H., Lisa Morrone Birkenthal, M.S., and Beth Balkite, M.S.

Committee News 'n Notes

Quality Assurance in Genetic Services — Can we Get There from Here?

by Karen Greendale, Genetic Services Program Administrator, New York State Department of Health

The time seems right for consideration of organized and well-thought-out methods for assuring quality in our genetic service centers. Although agencies such as CORN are expending considerable effort on issues relevant to quality assurance (QA) in genetic testing laboratories and the ABMG addresses issues relating to quality of genetic service providers, review of the scientific literature reveals surprisingly little discussion of QA in the clinical genetics arena. Several researchers have tried to evaluate genetic services by looking at patient satisfaction, post vs. pre-test knowledge or reproductive outcomes following genetic counseling. Although these data are relevant to QA, they do not address many crucial aspects. A major problem is the difficulty of defining the "outcomes" a quality program is supposed to achieve.

Applying general principles from the substantial literature on QA in clinical medicine to the particular spheres of clinical genetics and genetic counseling may prove to be illuminating. Modification of these principles to create model standards or peer review mechanisms could identify areas in need of improvement to assure patients and families the best possible quality of care.

An Example to Consider

As a first step, it may be important to explore how programs are currently monitoring quality. For example, all clinic conferences are not created equal. Some include various subspecialists from outside the genetics unit; others are limited to genetics staff. One program reviews every case; another presents a single interesting patient for educational purposes; a third group discuss only those cases which meet specific criteria; a fourth depends on informal discussion in the halls.

Opponents of formal QA might suggest that this sort of variation allows different programs to capitalize on their strengths; proponents would counter that variation without minimal standards is not in the best interest of patients. Such discussion will become more important as the explosion of new genetic information from the Human Genome Project brings more citizens into contact with genetic service providers. I would argue that personnel shortages at all levels will further stress the system, leading to increased hiring of non-traditionally trained colleagues and making QA even more important.

Other's Experiences

Numerous committees have called for standard-setting in this arena. ASHG's Human Genome Committee, in a letter to James Watson, Director of the NCHGR, states that: "The genome project must recognize at an early stage the potential for incorrect or inappropriate use of the new materials, by providing funds for...the development of national and international standards of practice and of standards for the quality control of diagnostic services, an area in which the human genetics community...will wish to play an important role."[1] ISONG has established a committee chaired by Judith Franklin, RN, to work on standards of practice for nurses providing genetic health care services. Various regional and statewide efforts are now underway. Particularly noteworthy are the activities of GLARGG and ongoing efforts in California, Texas, Tennessee and Florida.

NSGC Efforts

Ed Kloza has asked me to chair an *ad hoc* committee to work with colleagues from the clinical genetics, nursing and public health fields on these issues. Members of this committee are: Lisa Amacker North, Robin Blatt, Cam Knutson, Sarina Kopinsky, Brynn Levy, Ilana Mittman, Elsa Reich and Tillie Young.

This is a new idea for most of us. Like all controversial ideas, this one will benefit from considerable discussion. Committee members would be interested in hearing from anyone working on QA in the genetics setting or from colleagues who would like to share their points of view on this subject.

1 *Am J. Hum. Genet.* 49: 687-91, 1991.

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GUIDING PRINCIPLES AND RESOLUTIONS...

weapons. The NSGC's efforts to adopt such policies parallel these practices.

NSGC's Political Voice

As the number of NSGC members grows, we are able to increase our political voice and influence legislative and judicial activities that impact the health care services available to our patients. Formal statements endorsed by our membership strengthen our organization's political position. Members can speak not only from their personal beliefs, but with the authority of organizational support.

Guiding Principles and Resolutions become valuable when we are asked to support positions represented in various court cases. The NSGC often is requested to sign *amicus curiae briefs*, "friend of the court" documents containing arguments supporting one side of an issue. In some Supreme Court cases, hundreds of *amicus curiae briefs* are submitted, giving voice to those having an interest in the outcome. The NSGC is often called upon to align with other organizations, usually professional health groups, who have interests similar to ours. Our Reproductive Freedom Resolution allows the legislative liaison to sign *amicus curiae briefs* for specific cases regarding reproductive freedoms without needing a Board quorum each time.

Guiding Principles and Resolutions Serve as Public Belief Statements

Guiding Principles also provide definition for the NSGC, allowing members to speak with confidence on behalf of the organization about controversial issues.

Just as our Code of Ethics is a document for internal use, the Guiding Principles and Resolutions provide a mechanism for representation in external situations. In non-legislative activities, Guiding Principles allow members to represent issues of the Society to the media, to other professional organizations and to private industry. Guiding Principles and Resolutions allow *our own* organization to further define our policies instead of permitting the media or other professional groups to interpret our philosophy.

Status of Guiding Principles

During the Board of Directors meeting in October 1991, four Guiding Principles were passed. These statements, representing some beliefs universal to the practice of genetic counseling, have been discussed, revised and reviewed by many committee members and the Board. The Guiding Principles include statements covering: access to care, non-discrimination, confidentiality of test results and disclosure and informed consent. *(See box below.)*

Resolutions Differ in Timeliness and Temperature

In contrast to the more universal nature of the Guiding Principles, Resolutions are timely, and they may be temporal and change as laws or available services change. The Legislative Subcommittee, chaired by Trish Magyari, has studied and prioritized many topical issues pertinent to our Society. These issues have resulted in the two resolutions, Prenatal Substance Use and Fetal Tissue for Research, currently proposed to the membership by the Social Issues Committee.

Proposed Resolution: Prenatal Substance Use

In the past few years, national attention has focused on women who abuse alcohol and drugs during pregnancy. Several states have tried to prosecute these women. These same states have limited treatment services for pregnant women's addictions. Treatment services have proven to be a successful way in which to overcome both drug and alcohol abuse. Therefore, the proposed Resolution is:

> *"The NSGC supports increasing prevention efforts and treatment services for alcohol and drug dependent women and their children. These services are*

Guiding Principles

Access to Care: The NSGC supports individual access to appropriate genetic services regardless of racial or ethnic background, socioeconomic status, disability, ability to pay for services or method of payment. Access to care for families with genetic concerns is also necessary in the areas of prenatal care, family planning services, pediatric care and psychological counseling. *(Adopted 1991)*

Nondiscrimination: The NSGC opposes discrimination against an individual with regard to eligibility for or maintenance of employment, insurance coverage or medical benefits on the basis of the results of genetic testing. Consideration of testing information is appropriate only when used to protect the individual's best interests. *(Adopted 1991)*

Confidentiality of Test Results: The NSGC supports individual confidentiality regarding results of genetic testing. It is the right and responsibility of the individual to determine who shall have access to medical information, particularly results of testing for genetic conditions. *(Adopted 1991)*

Disclosure and Informed Consent: The NSGC supports an individual's right to full disclosure of all appropriate medical options regarding reproductive testing and management of genetic diseases and birth defects. It is the care provider's responsibility to provide effective communication of all available options and to obtain informed consent for procedures involving risk to the individual or fetus. *(Adopted 1991)*

Resolution

Reproductive Freedom: 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. *(Adopted 1987)*

...EXPLAINED from p. 1

far preferable to punitive sanctions brought against alcoholic and drug dependent women solely because they were pregnant when they used alcohol or drugs."

Proposed Resolution: Fetal Tissue Research

Fetal tissue research has created a great deal of controversy in recent years. In 1988, the Federal government banned research funding for fetal tissue transplantation. Subsequent efforts to regulate this area of research have been unsuccessful. In September 1991, an ACOG news release announced a "New Board to Monitor Pre-embryo and Fetal Tissue Research." This board, comprised of physicians, lawyers, ethicists, scientists and religious leaders, will set guidelines and review ethical issues in reproductive and fetal tissue research and fetal tissue transplantation. The hope is that this advisory board will benefit patients, physicians and scientists alike. The NSGC's proposed Resolution on Fetal tissue research reads as follows:

"The NSGC supports fetal tissue research (within strict medical guidelines) as a legitimate and important area of scientific investigation and as a vital avenue of research toward treatment of genetic conditions."

NEXT STEP BELONGS TO THE MEMBERSHIP

The Social Issues Committee has dedicated itself to making social policy recommendations to the NSGC Board and membership.

The Board has approved the Resolutions, and voted that they be presented to the NSGC membership for review, discussion and vote. Contact your Regional Representative prior to the May 3 Board meeting or write a Letter to the Editor for the next issue of *PGC*. Pending legal review, a postcard enclosed in the next issue of *PGC* will provide you with the opportunity to vote on these recommended organizational Resolutions.

COMMENTARY FROM MEMBERS OF SOCIAL ISSUES COMMITTEE

PRENATAL SUBSTANCE ABUSE... UPHOLDING OUR COMMITMENT TO HELP WOMEN IN NEED OF INFORMATION AND HEALTH CARE

by Trish Magyari, M.S., Macro International, Inc., Silver Spring, MD

The Resolution on Prenatal Substance Use was originally proposed to the Board in 1988 in direct response to requests from other professional organizations for our position on this issue. The Coalition on Alcohol and Drug Dependent Women and Their Children and the National Perinatal Association, of which we are members, subsequently passed similar proposals. Since that time, other organizations, including the American Public Health Association, the AMA and the American Nurses Association, have adopted "non-prosecution" policies for addicted women.

The fact that other organizations have passed a similar policy is supportive, but not the primary reason the NSGC is considering this Resolution. Rather, it stems from the Guiding *Principles* of Access to Care and Non-Discrimination, as well as our professional commitment to provide services to women and children in need of health care. The need for additional alcohol and drug treatment programs for pregnant women and mothers is well documented. Additional efforts aimed at *preventing* alcohol and other drug related birth defects, a charge of many genetic counselors, are also needed. However, despite this, and the fact that alcoholism and drug addiction are diseases amenable to treatment, women have been prosecuted and jailed in several states *solely* because they were pregnant when they used alcohol or drugs. The fear of prosecution or mandatory reporting to state agencies is driving the very women most in need of services out of the health care system. Moreover, the trend to apply punitive measures in a disproportionate manner to low-income women and women of color raises serious concerns about discrimination.

FETAL TISSUE RESEARCH... FACE OFF BETWEEN CONFLICTING ETHICS

by Deborah Durand, M.S., Genetics Institute of Florida, West Palm Beach

Currently in the U.S., centers may not use federal funding for fetal tissue research or organ transplantation. A change in legislation cannot occur until the ethical dilemma of using fetal tissue or organs is resolved.

Proponents contend that fetal tissue is less differentiated and immunologically reactive than adult tissue, thereby more adaptable for use in transplants. This offers hope to patients suffering from advanced stages of degenerative disorders, Parkinson's and Alzheimer's disease among them. They argue that we have a responsibility to those individuals whose health might be restored. Transplanting organs from neonates with lethal disorders to save other infants suffering from organ failure is another benefit. To some parents who have a baby with an isolated neural tube defect, such as anencephaly, the idea that organs from their nonviable neonate may benefit another infant's life is comforting. Currently, those parents are denied that small amount of solace because opportunities are not available.

Opponents of fetal tissue research argue that abortion is so morally wrong that the end does not justify the means. Many worry that if we allow research or clinical use of fetal tissue or organs, women may terminate their otherwise normal fetus to enhance someone else's life. That argument would not apply in the case of anencephalic fetuses or neonates. However, they feel that situations would exist in which parents would deny life in an attempt to postpone an inevitable early death. Additionally, they express concern about setting a precedent for further, more morally repugnant behaviors.

DIAGNOSTIC TESTING FOR FRAGILE X SYNDROME *from p. 1*

penetrant males, and fetuses at risk for mental impairment. At present, prenatal cytogenetic studies should be performed to complement direct DNA studies. In certain cases when the direct method is inconclusive, linkage studies may be necessary.

DNA TO REPLACE CYTOGENETICS?

With issues regarding the usefulness of direct DNA analysis for carrier and prenatal testing somewhat resolved, the question remains whether DNA analysis should replace cytogenetic studies as the primary diagnostic tool for detecting fragile X syndrome in mentally retarded or learning disabled at-risk individuals.

Some researchers feel strongly that cytogenetic studies should be the primary fragile X diagnostic tool. Although most agree cytogenetics will eventually be replaced by DNA studies, experience with DNA mutation analysis is still limited. Therefore, some people fear potential DNA diagnostic problems may not have been realized.

Difficulties include interpretation, especially when diagnosing in a young child. For example, repeat size cutoff points for distinguishing normal polymorphisms from premutations and premutations from full mutations have not yet been clearly defined. There is also a small number of individuals who demonstrate the fragile X chromosome and are phenotypically fragile X, but fail to show the fragile X mutation.

The standard laboratory test available has been a laborious, specialized cytogenetic study that induces the fragile X site. The accuracy of this testing is approximately 99% in intellectually impaired males and approximately 90% in affected females. Clinical interpretation is required on low positives (less than 4%) which may or may not represent false positives. DNA direct diagnosis may prove more reliable than cytogenetic studies. Some individuals who test negative on cytogenetic studies have been positive for the DNA fragile X mutation. Percent fragility has not been shown to correlate with intellectual involvement in fragile X males or females. On the other hand, there is a relationship between increased length of inserted DNA fragment and degree of mental impairment. There-fore, DNA mutation analysis should be of greater value in predicting mental status. DNA mutation analysis is less tedious, faster, and less expensive. With the recent development of a PCR method that amplifies across the full mutation, molecular testing may be performed even more efficiently.

There are pros and cons for each diagnostic technique. The ordering protocol depends on several factors. As always, the strengths and limitations of the cytogenetic and molecular laboratory should be scrutinized. A center's experience, choice of cytogenetic culture methods or DNA probes, turn around time, use of controls and even research endeavors should all influence our final decision. For the patient, financial considerations, including what method third party insurance will reimburse, may be important. Cytogenetic studies (which usually include routine chromosome analysis) range from $475 to $800, while DNA ranges from $180 to $350.

REASONS FOR STUDIES

If direct DNA analysis is pursued as the first diagnostic step and the patient does not show the mutation, it is critical to perform high resolution chromosome analysis to rule out other chromosomal explanations. For patients highly suspect for fragile X syndrome, you also may consider fragile X cytogenetics to rule out the subset of individuals who are cytogenetically positive but mutation negative. For those who choose to stick with cytogenetics as the primary test, mutation analysis may be warranted for cytogenetically negative patients who raise a high degree of suspicion for fragile X. DNA mutation analysis should be used to confirm the diagnosis in low cytogenetic expressors.

From a patient perspective, the new DNA testing has had the most impact on intellectually normal relatives who had been concerned about the accuracy of previous carrier detection methods or who may have had inconclusive results. Parents of children diagnosed with fragile X welcome the new testing since it has the potential to determine carrier status in their normal functioning children. The predominant hope for patients, however, is that the discovery of the FMR-1 gene will lead to a clear understanding of the protein, or lack of protein, responsible for their child's intellectual impairment. Medical therapy is currently not available, but molecular advances have rekindled hope for the future.

Our understanding of the fragile X gene has improved dramatically in the last year. The diagnostic methodology will continue to change and recommendations for at-risk families will be modified. Informed counselors must take an active role in this discussion since they will often be responsible for explaining the changes to the at-risk families.

SUGGESTED READINGS

Fu U et al (1991): Variation in the CGS repeat at the fragile site. Results in Genetic Instability: Resolution of the Sherman Paradox. *Cell* 67:1047-1058.

Pergolizzi RG et al (1992): Detection of full fragile X mutation. *Lancet* 339-271-272.

Rousseau F et al (1991): Direct diagnosis by DNA analysis of the fragile X syndrome of mental retardation. *N Engl J Med* 325:1673-1681.

Shapiro LR (1991): The fragile X syndrome. N Engl J Med 325-1736-1737.

RELATED CONFERENCE

The National Fragile X Foundation's 3rd International Conference will be in Snowmass/ Aspen, June 16-20. The key topic will be the molecular aspects of the fragile X syndrome, including the most recent research endeavors and their clinical applications. For further information, contact the National Fragile X Foundation at 1-800-688-8765.

ABMS Report...Evaluating the Counselors' Status

The American Board of Medical Specialties (ABMS) has recently accepted as its newest member the American Board of Medical Genetics (ABMG). As its twenty-fourth member, the ABMG will represent Medical Genetics interests to the American Medical Association. As a condition of membership, however, the ABMG "will not certify Master's Degree level genetic counselors."

A letter was included in our membership mailing last fall and Anne Spence, PhD, President of the ABMG, spoke directly to our membership about these impending events at our membership meeting in Washington. She assured genetic counselors that if the ABMG became part of the ABMS, the ABMG would continue to support counselors' interests.

Basic Questions, Issues, Concerns to be Addressed

How then can the ABMG agree not to certify Master's level counselors and yet maintain them as members? This question is one that is being addressed by a working group of genetics professionals on March 10 in Philadelphia before the scheduled March 12 ABMG Board meeting. Anne Spence, Charles Epstein, Diane Baker and Debra Collins will represent the ABMG at this meeting, while Betsy Gettig, Joan Scott, Ann Walker, Ann C.M. Smith and I will represent the NSGC. Attorneys for both organizations will also be in attendance.

The ABMG will clearly need to be restructured as a result of its membership in the ABMS. The group will address the issues of preservation of certification status and value, representation on appropriate governing bodies and continuation of examinations. Sure to be on the agenda will be institutional accreditation as well.

Ongoing Communications

Because the ABMG by-laws require a two-thirds vote for amendment, the ABMG membership will be called upon to approve this restructuring. If a restructuring proposal that protects Master's level counselors can be negotiated, I will ask the NSGC Board of Directors to review it. Our Board may choose to endorse or reject any proposal. The NSGC membership will be made aware of the NSGC's Board position.

But it is the ABMG membership, not the NSGC, that will vote on the restructuring proposals.

Since Master's level genetic counselors constitute thirty-nine percent of the ABMG membership, it would appear that any successful proposal will need to be supported by the NSGC membership.

Impact, Action Must be Weighed

These events have more potential impact on the future of the profession of genetic counseling than any situation with which the NSGC has previously dealt. Regional Representatives will be informed as events unfold, as will all Board members. NSGC members are asked not to take any action until the Board has had an opportunity to review and discuss all options and make a recommendation to the membership. At that time we would expect and encourage reasoned discussion.

Edward M. Kloza, M.S.,
President,
March 6, 1992

Human Genome Project Update

This column will regularly provide an update of information about the Human Genome Project. All opinions, ideas and information for the column are welcome. — JoAnn Inserra, Norwalk Hospital, Norwalk, CT

CF Pilot Projects

The researchers who received NIH funding for clinical studies of testing, education and counseling for cystic fibrosis mutations include: Joanna Fanos, Children's Hospital Oakland Research Institute, Oakland, CA, Wayne Grody, UCLA School of Medicine, Los Angeles, CA, Neil Holtzman, Johns Hopkins University, Baltimore, MD, John Phillips, Vanderbilt University, Nashville, TN, Peter Rowley, University of Rochester, Rochester, NY, and James Sorenson, University of North Carolina, Chapel Hill, NC.

Genetics Resource for African American Families

Researchers at Howard University are expanding the university's human genome research program by organizing an African-American reference family panel. This will be a collection of family histories and DNA samples from African-American families. The information will help scientists identify gene-based differences in drug responses and susceptibility to diseases and environmental factors among different population groups.

Genome Data Base

NIH and DOE have awarded Johns Hopkins University School of Medicine $5.3 million to support the Genome Data Base, a repository for information about the location and description of genes including disease genes and other genetic markers on chromosomes. Hopefully, this will ensure worldwide access to accurate information for human genome research.

Education Tools

- ***Booklet:*** "The HGP: New Tools for Tomorrow's Health Research." Describes the background behind the HGP and includes a discussion of goals. It is geared to the non-scientist. Multiple copies (up to 50) are available from Leslie Fink at the NIH office of communications (301-402-0911)

What's Ahead?

- ***May 6-10.*** Genome Mapping and Sequencing Workshop (GDB exhibit displayed), Cold Spring Harbor, NY
- ***May 21.*** The HGP and the Future of Medicine; Bethesda, MD (C. Dahl, 301-402-0338).

SPF Awardee Named

Beverly Tenenholz, West Penn Hospital, Pittsburgh, is the first recipient of the NSGC's Special Projects Fund (SPF). The proposal, "Now that You've Been Told Your Baby has..." is to develop a series of educational brochures for use by patients who elect to continue a pregnancy in which fetal abnormalities are prenatally detected. While some couples choose abortion when faced with knowledge that their baby will have a birth defect, others elect to continue the pregnancy. Available patient literature often does not address prenatal concerns such as delivery options and neonatal treatment for these couples.

Four booklets for couples told that their baby has spina bifida, Down syndrome, hydrocephaly or congenital heart disease will be developed. Each booklet will have six sections dealing with issues such as reaction to the diagnosis; perinatal decisions about pregnancy care; intervention after delivery; and facing future long term issues.

The SPF was established as a result of the NSGC's 10-Year Anniversary Project fund drive conducted in 1988. According to the guidelines, interest on the original amount raised will be awarded for projects that focus on the future of the genetic counseling profession and/or the provision of genetic services. Applicants must be members in good standing of the NSGC. Policies and procedures for applications are on p. 19 of *Toward the Future,* an NSGC publication distributed at the International Congress of Human Genetics, or they can be obtained by contacting the NSGC Executive Office.

Application deadline is May 15.

Collins Awarded HGP Grant

Debra Collins, University of Kansas Medical Center, has received a grant entitled "Human Genetics Education for Middle and Secondary Science Teachers" from the Department of Energy. The goal of the 3-year, $600,000 grant is to address the lack of public information on the Human Genome Project (HGP) through educational workshops for science teachers on the social, legal and ethical issues of the HGP.

Teachers will be recruited for a four phase national program to prepare them to become "resource" teachers, selected for their knowledge, experience and links with existing teacher organizations. Workshops will be conducted to update and expand the use of human genetics curricula materials using an inquiry orientation and hands-on materials. Genetic counselors will be recruited to serve as mentors as teachers revise their curricula to incorporate the HGP.

Kudos to New Journal

Congratulations to Editor **Deborah Eunpu** and the editorial board, **Joan FitzGerald, Rita Beck Black, Janice Edwards, Beth Fine, Seymour Kessler, Edward Kloza, Mark Lubinsky, Anne Matthews, Bradley Popovich, Robert Resta, Susan Schmerler** and **Ann C.M. Smith** as well as the authors of the inaugural issue of the *Journal of Genetic Counseling.* If the first issue is an example, this will be a wonderful forum to enhance and enrich our profession. Consider submitting your ideas and receive assistance from the Editorial Board.

Members in good standing who have not yet received their copy may contact Human Sciences Press, Inc. 233 Spring Street, New York, NY 10013-1578.

[Note: Since the Journal is posted at bulk mailing rates, it is particularly critical to maintain accurate address information with the Executive Office. The postal service does not forward bulk mail.]

Genetic Storytelling Panel Features Members

Luba Djurdjinovic, Beth Fine and **June Peters** will present a workshop, "Eliciting the Family Story: Implications for Genetic Counseling" at a conference entitled *Medicine and Its Stories* at the Annual Conference of the Society for Health and Human Values. The four-day conference will be held in Tampa this spring.

The presentation will focus on illustrating how histories in genetic counseling sessions impact the family or individual's ability to cope with a genetic disease or risk, based on careful attention to psychosocial, marriage and family, ethnocultural and religious views.

Congratulations to Luba, Beth and June for bringing the genetic counseling perspective of storytelling to the attention of a new audience of healthcare professionals, policy developers, scholars, teachers, clergy and lawyers.

Code of Ethics Named to Honor Roll

NSGC's Code of Ethics has been acknowledged on the American Society of Association Executives' (ASAE) Associations Advance America Awards Honor Roll. President Bill Taylor commended the NSGC's Code and the valuable contribution it makes to society at large. The award application was submitted by Executive Director **Bea Leopold** and Code of Ethics subcommittee chair, **Judith Benkendorf.**

Region III Counselors Active in SERGG Activities

Stephanie Smith, University of Mississippi Medical Center, Jacksonville, has been elected to represented SERGG on the CORN Education Committee. Long active in Region III activities, she has co-chaired the SERGG Genetic Counselors Workshop for the past several years with **Ron Cadle.** Stephanie and Ron have agreed to co-chair the 1993 NSGC Annual Education Conference. Both have served in recent years as Region III Representatives to the NSGC Board.

Some of the ideas of our members can stimulate ongoing discussion, others demand immediate rebuttal. To that end, this forum will provide either a Viewpoint or a Point/CounterPoint. This Viewpoint, an idea for deliberation, was suggested by Sue Schmerler. We took advantage of the expertise of one of our members who has previously co-authored a prominent publication on this subject. As always, we invite responses and suggestions for future columns.

Seth Marcus, M.S.
Editor, Viewpoints/Point-Counterpoint

ADVANCED PATERNAL AGE: RISK AND REASON

by Elsa Reich, M.S., Human Genetics Program in Pediatrics, New York University School of Medicine

The effect of paternal age on the frequency of birth defects has been recognized since Penrose[1] observed that the age of the fathers of children with achondroplasia was increased over the median age of fathers in the general population. This observation has been confirmed in numerous studies of other dominantly inherited conditions. Likewise, grandfathers of boys with X-linked conditions have also been noted to have ages greater than that of control populations. Although studies done in the 1970's showed an increased frequency of chromosome abnormalities in the children of older fathers, more recent data have failed to confirm this association.

Should genetic counselors inform patients about this risk? If they should, when should they do so?

WHAT IS THE RISK?

The magnitude of the risk is unclear. Friedman[2] suggests that the chance for a 40 year old man to father a child with a condition caused by a new dominant mutation is at least .3-.5%, and roughly one-third of all babies with these conditions are born to fathers in this age group. These estimates may be biased since the author assumed that the rate of mutation is similar in all dominantly inherited conditions. Risch *et al*[3] suggested that this was not the case and that rates of mutations differed. The total risk to the individual may not be easy to calculate.

Many dominantly inherited conditions occurring anew cannot be detected prenatally. Should we inform couples in which there is an "older" father during a preamniocentesis/CVS session that they have an unknown risk to have a child with a dominantly inherited condition and that we most likely will not be able to detect it since there are over 1000 such diseases?

The total risk to the individual may not be easy to calculate.

The level of anxiety that pregnant couples experience is increasing. Should we risk increasing anxiety with uncertain data? While I do not believe in withholding information from patients, the amount of information that patients must consider and understand is growing, and we must carefully weigh the importance of each piece of information and its use to the patient. If information about the effect of paternal age is requested, the data should be presented as a small, but undefined increased risk. If information is *not* requested, I do not volunteer the data, since the total risk to the pregnancy is not altered dramatically and little is available to ameliorate the risks. Exceptions to this practice may occur when the father is in his sixties or seventies.

SHARING THE RISK?

Some people feel that the "older" woman bears too much responsibility for the increased frequency of abnormalities, and this "responsibility" should be shared more evenly by informing couples of the risks associated with the father's age. After all, Friedman estimates that these risks are similar to those of a 35 year old woman having a baby with a chromosome abnormality.

One might consider routinely presenting the information in a relatively non-threatening way by referring to it at the same time that one discusses the general population risks with a comment that it does not change the "range" of risks significantly. One can alert the couple to the risks, but not attach an undue significance to it. Even those counselors who do not routinely discuss paternal age-related risks may wish to do so when too much emphasis is placed on the maternal age-related risks. It may be useful to diffuse the mother's sense of responsibility by commenting on the paternally associated risks. Sometimes a bit of humor is called for at a time such as that.

There are some circumstances when these data may be communicated routinely, in contrast to the pre-amniocentesis/CVS session. Couples coming for preconception counseling to understand their reproductive risks should be apprised of all of their risks. It is a more appropriate time to discuss, along with the general population rates of abnormalities, the risks associated with both the maternal and the paternal ages.

1 Penrose, L.S. 1957. Parental age in achondroplasia and mongolism. *Am J Hum Genet* 9:167.
2 Friedman, J.M. 1981. Genetic Disease in the offspring of older fathers. *Ob Gyn* 57:745.
3 Risch, N., Reich, E.W., Wishnick, M.M., McCarthy, J.G. 1987. Spontaneous mutation and parental age in humans. *Am J Hum Genet* 41:218.

Bulletin Board

ACPCA Conference in Portland

The 49th Annual Meeting of the American Cleft Palate-Craniofacial Association will be held in Portland, Oregon from May 13 - 16. Two symposia will be presented prior to the meeting: "Cleft and Craniofacial Patient Management During the First Year of Life: Starting Off on the Right Foot!" will be held on May 11 and 12. "Making a Difference: Nurse Involvement in Pre-Operative Teaching, Post Operative Care and Follow-Up" will be held on May 12.

For information, contact Nancy C. Smythe, Executive Director, ACPA, 1218 Grandview Ave, Pittsburgh, PA 15211; 412-481-1376.

March of Dimes Conference

The March of Dimes Birth Defects Foundation will hold its 24th Annual Clinical Genetics Conference, "Clinical and Molecular Cytogenetics of Developmental Disorders," at Stanford University School of Medicine, July 12 - 15.

For more information, contact Carol Blagdowidow, 914-997-4524.

HHS Position Paper Available

"Fetal Alcohol Syndrome and Women Who Abuse Alcohol: An Overview of the Issue and the Federal Response," by Barbara Anderson and Emily Novick, Program Analysts, is now available and can be obtained through the US Dept HHS, 202-245-1880.

Feds to Study QA in Genetics

A congressional subcommittee is gathering information on:

- Patients who were given mistaken information by their physicians;
- Physicians who were given misinformation about test results from genetic testing laboratories;
- Any patient problems arising from misinterpretation of test results, misrepresentation of risks or management options by physicians or genetic testing laboratories.

The subcommittee hopes to find ways to maintain high standards of quality in the genetic testing area. Any counselor who wants to discuss a case should contact: Dr. Charles A Gardner, Subcommittee on Human Resources and Intergovernmental Relations, Government Operations Committee, B-372 Rayburn House Office Bldg., Washington, DC 20515; 202-225-2548; FAX# 202-225-2382.

Report on Membership Poll

Of the 1011 postcards included with the last issue of *PGC*, 379 (37%) were returned. Of those responding, 181 (48%) indicated that "The NSGC should hold its 1993 AEC in New Orleans" and 196 (52%) indicated that "The NSGC should not hold its 1993 AEC in New Orleans."

GC Doctoral Track Offered

The University of Pittsburgh's Department of Human Genetics invites inquiries about a new track leading to a Ph.D. in Human Genetics (Genetic Counseling), designed to prepare candidates to pursue research in genetic counseling. Candidates should be board certified, have a minimum of three years experience and be committed to a research career.

Inquiries can be directed to: John J. Mulvihill, M.D., Dept. Human Genetics, Graduate School of Public Health, University of Pittsburgh, 130 DeSoto St, Pittsburgh, PA 15261; 412-624-9951; FAX# 412-624-3020.

Directory Errors Corrected

Our apologies to the following members who reported errors in the 1991-92 Membership Directory:

Berliner, Janice: correct FAX# is 908-526-2400x2534

Blatt, Robin J.R.: correct address is Mass. Genetics Program (delete Central Library FHS, Mass. Dept. Public Health)

Fishbach, Andrea: correct address is Kaiser Permenente, Genetics, 2280 Geary Blvd, San Francisco, CA 94115; 415-202-2993

Jensen, Karen: correction to read Dept Communications *Disorders*

Krivchenia, Eric: correct FAX# is 313-993-0153

Marini, Tina: correct hospital name is Bay*state*

Markel, Dorene Samuels: correct phone # is 313-764-8050; correct FAX# is 313-764-2189

Mitter, Navnit: correct company name is SmithKline *Beecham* Laboratories

Shouse, Dolores: correct as follows: 1120 Maricopa Hwy, Ojai, CA 93023; 805-646-5555

Walsh-Vockley, Catherine: correct phone # is 507-284-2306, correct FAX# is 507-284-0161.

Please note these changes in your directories.

Research Network

Emotional Responses to CVS

Pregnant women planning to undergo CVS are being sought for a doctoral candidate's dissertation. Eligible subjects must meet the following criteria:

- Previously borne a child with a trisomy chromosomal abnormality
- age 34 or less
- no prior experience with either amnio or CVS
- no history of a chronic physical or mental disorder

Subjects will be asked to complete a brief, standardized questionnaire and a demographic data form prior to undergoing CVS. The entire process will require about 10 minutes. Counselors wanting more information, or who have qualifying patients, are encouraged to contact: Suzanne Zamerowski, MS, 336 Robin Hood Dr., Yardley, PA 19067; 215-295-5286 (collect).

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Hirschsprung Disease Study Seeks Family Data

A molecular genetic study of Hirschsprung disease is being conducted at the University of Pittsburgh, Department of Human Genetics. Two types of families are eligible to participate:

- Those with more than one living member affected
- Those with individuals with multiple abnormalities *and* Hirschsprung disease, excluding cases with Down syndrome and Hirschsprung disease.

Inquiries and response can be directed to: Dr. Aravinda Chakravarti, GHPH, Dept Human Genetics, University Pittsburgh, 130 Desoto St, Pittsburgh, PA 15216; 412-624-3066.

COMMITTEE NEWS 'N NOTES

NOMINATING COMMITTEE

NEW DEADLINE OFFERS 2ND CHANCE

The Nominating Committee has extended by two weeks the deadline for nominations. Consider nominating yourself or a colleague for Board leadership in the following capacities: President-Elect, Secretary and Representatives to Regions II, IV and VI.

Nominations may be sent to Roseann DiMaggio, Integrated Genetics, One Mountain Road, Framingham, MA 01701, and must be postmarked *no later than April 3.*

Members of the 1992/93 nominating committee are: Joan A. Scott, Chair; Roseann DiMaggio, Susan Schmerler, Bonnie LeRoy and Kathleen O'Connor.

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LONG RANGE PLANNING

VISION TO BE PRESENTED

The Long Range Planning Committee has developed a draft vision for the NSGC based on the focus groups held at the International Congress of Human Genetics and feedback of members.

The draft will be presented to the Board at the interim meeting in May and will be presented for membership discussion at an open forum during the 1992 Annual Education Conference in San Francisco.

Upon Board and membership approval, the next phase of developing a Strategic Plan will begin. For more about the process, contact committee members: Ginny Corson, Chair, Debra Collins, Andrea Fishbach, Denise Greene, Ann Happ Boldt, Trish Magyari, Mimi Riesch-Donnelly or Bea Leopold.

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EDUCATION COMMITTEE

AN OPPORTUNITY TO EDUCATE

BSCS and the AMA have created a module about the Human Genome Project (HGP) for use in high school biology classes with a grant from the Department of Energy. This program highlights the scientific and technological advances made in the field of human genetics, some of the ethical issues raised by the HGP, and the development of public policy. The module has been reviewed by members of the NSGC, ASHG and CORN as well as others. It currently is being field-tested by high school biology teachers.

If you work with high school teachers, or have thought about expanding your educational efforts, consider the following:

- Let the biology teachers in your community know that a free copy of the module will be mailed to more than 50,000 instructors nationwide by mid October.
- Encourage those teachers who do not receive the module to contact BSCS, 830 N. Tejon St, Suite 405, Colorado Springs, CO 80903.
- Contact your State Biology Teachers Association to present a topic related to the HGP.
- Prepare information packets about cystic fibrosis and/or Huntington disease, disorders highlighted in the students' activities.

Copies of the module and other education ideas will be available at the AEC in San Francisco.

NSGC CONSIDERS SHORT COURSES

Our careers continuously challenge us to remain current. The Annual Education Conference and regional meetings serve this purpose, but cannot provide an in-depth program about one topic.

Many professional organizations provide refresher courses or short educational programs for their members. These courses serve many purposes, from updating new information or research to preparing for a career change within the profession. NSGC's Education Committee is investigating the interest and need for intensive workshops or "short courses" for genetic counselors. Topics can range from science, technology and new counseling methods to writing, administration and business skills.

If you would like the NSGC Education Committee to establish a program of "short courses," please complete and return the enclosed postcard. We encourage you to discuss this potential membership service with your colleagues and urge them to complete the postcard. If the NSGC membership shows little support for this idea, no further action will be taken.

ANNUAL EDUCATION CONFERENCE

THE HUMAN GENOME PROJECT: IMPACT, IMPLICATIONS AND ISSUES

(OR WHEN WE'VE SOLVED THE MYSTERIES, WHAT WILL WE DO WITH THE CLUES?)

DATES:	November 6 - 8, 1992
LOCATION:	Grand Hyatt on Union Square, San Francisco, CA
CHAIRPERSONS:	Ann Happ Boldt and Lynn Hauck
NEW FEATURES:	Nine workshops in three time blocks, featuring these hot topics: quality assurance, DNA technologies, applications of Code of Ethics, difficult dilemmas*, social, ethical & legal perspective of the HGP, support groups, private practice, psychotherapeutic perspectives in genetic counseling, and student issues
NETWORKING OPPORTUNITIES:	Private Practice • Beth Balkite (203-431-0537) Cancer Genetics • Maureen Smith-Deichmann (312-702-0681)
BACK BY POPULAR DEMAND:	Open Forums
REGISTRATION INFORMATION:	To Be Mailed in April

** Difficult Dilemmas (formerly Curbside Consultations) has been incorporated into workshop format. Volunteers are needed to present a 10-minute case summary focusing on, but not limited to,, the following issues: confidentiality, ethical decision-making, ambiguous prenatal test results, family dynamics, adoption, pediatric HIV diagnoses, counseling the mentally ill or paternity identification. To discuss submissions, contact Kathleen Delp [517-355-2724] or Lavanya Misra [212-523-4474].*

GENE BYTES

A Major Improvement over Typewriters

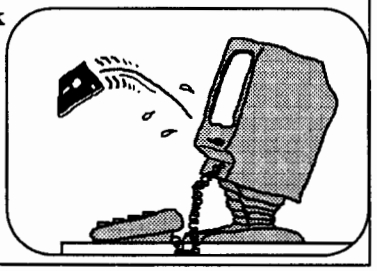
by Karen Wcislo, MS and Robert Resta, MS

Word processing (WP) is not simply new and improved typing. Good WP software offers helpful tools to enhance some of the tasks of the genetic counselor. Whether you already own or are thinking about buying WP software, consider the following:

- **Tables.** Need to make an Age/Chromosome Risk Chart? Prepare a summary table for a presentation? Your software should allow you to define a table of virtually any size as well as to define the appearance and thickness of lines. The software should also be able to import data directly from a spreadsheet into the table, a time-saving feature when preparing annual clinic reports.
- **Redlining/Strikeout.** Working on a publication for the *Journal of Genetic Counseling?* A pamphlet with several authors? These features allow you to insert changes side by side or on top of the original text. This way, the senior author can track the original text along with suggested changes. In addition, a comments feature permits other authors to make comments which appear on the screen but are not printed.
- **Macros/Merge.** Macros record keystrokes, so frequently used commands or text can be recalled with 2 or 3 keystrokes. This is especially helpful when preparing letters with little text variation, such as a basic discussion of chromosomes or recessive inheritance. Using boiler-plate text called up by macros and individualizing the appropriate parts of the letter, one can quickly create patient follow-up letters. Merges will automatically insert text such as addresses or appointment times into otherwise standard letters. This is helpful when preparing many letters at once, such as appointment or result letters.
- **Graphics.** Do you prepare simple newsletters? Overheads? Graphic images help catch the audience's eye. Look for WP software that comes with some images and also allows you to import graphics from other software. Admittedly, this feature works better in Windows WP programs, but DOS programs will meet most of your needs.
- **Outlining.** Preparing a lecture or presentation? An outlining function can save a tremendous amount of time. Numbers, letters and indentations are automatically generated and updated for each section of the outline. Different numbering styles are available, or you can create your own.

Most of the sophisticated WP packages, such as Word Perfect and Microsoft Word, offer the above-described options. To many of us, learning new software evokes images of Marlow's river journey into the heart of darkness. However, you needn't master the entire software — just learn the features that make your job easier.

Bright Ideas...Do you use Word Perfect? Think about subscribing to Word Perfect Magazine ($18/yr). It offers information for beginners as well as pros.



Is there a software product you would like us to review? Send us the name of the product and company, and we will try to review it in future columns.

✍ LETTERS...

We are Not Ready for CF Screening

To the Editor:

We feel it necessary to respond to the two articles about CF carrier testing. (*PGC*, 13:4) Both articles in "ViewPoint" advocated CF screening of Caucasian pregnant women without a positive family history of CF. The thoughts expressed may be assumed by others to represent the views of our profession and perceived as standard of care.

CF carrier screening for couples without a family history is contrary to both the ASHG statement and NIH guidelines. While some counselors may feel that *any* detection is better than none at all, CF does not fit the basic criteria for a mass screening program. The genetics community learned valuable lessons regarding those criteria during previous experiences with carrier screening for Tay-Sachs and sickle cell disease. In order for a population screening program to be cost effective and properly implemented, the following factors need to be considered:

- severity of the disease,
- potential for treatment
- cost, accuracy, and sensitivity of the laboratory analysis and
- educational materials necessary to minimize potential psychosocial ramifications.

CF currently does not meet these established screening criteria.

There is increasing pressure to offer CF carrier screening. Our center recently confronted this issue and considered the following points (based on 40% participation of 25 CVS patients/week):

- One at-risk couple would be identified every 2 to 3 years.
- Three couples with one carrier member would be identified each month. This couple's risk will be increased without the availability of a definitive prenatal test.

We also struggled with how we would incorporate the necessary

information regarding the limitations of CF carrier testing into an already fact-filled and often anxiety provoking counseling session. The educational component presents an enormous challenge if there is to be true informed consent. This critical issue has not yet been fully addressed and is presently the focus of seven CF pilot programs.

Given these statistics and the significant amount of time necessary to explain carrier testing, we are currently not convinced that population screening of pregnant couples is appropriate.

The existence of a test does not obligate us to offer it. We need to be even more cautious if we have not even begun to understand the ramifications such a technology will have on society. As genetic counselors, we must take responsibility to ensure that proper guidelines, educational resources, and pilot programs are completed before mass screening efforts are undertaken.

Anne Greb, MS,
Eric L Krivchenia, MS,
Wendy R. Uhlmann, MS,
Joan V. Conard, MS,
Dana Arndt, MS,
Div. Reproductive Genetics,
Hutzel Hospital/Wayne State University, Detroit, MI

Go and Educate

To the Editor:

As a new member of the NSCG, I have been asked to cast a vote for or against holding our 1993 annual meeting in New Orleans. I have also read the arguments in favor of relocating the meeting to protest the legislative actions of the state's government. I believe that relocation is the wrong decision.

I am a native of the South, raised in the "good 'ole boy" politics of Texas, and it would be an understatement to say that these boys play hardball. Can we realistically expect to win this game if we don't even show up to bat? Do we step up to the plate in New Orleans, or forfeit the game entirely?

If we look to the Civil Rights leaders of the 50's and 60's for inspiration, we recall their names and faces, we recite their words, and *most of all*, we remember their presence in the face of adversity.

There is a silent majority in Louisiana that supports a woman's right to control her reproduction. The economic impact of relocation will be negligible compared to the message we could send to the legislature and the Pro-Life PACs they represent.

Not only can we send a message that we are unwilling to compromise our freedom of choice, we can use our presence in New Orleans to educate and empower the citizens of Louisiana. Moreover, our Code of Ethics challenges us to "Participate in activities necessary to bring about socially responsible change."

I believe that the only way we can make a difference is to play ball!

Jennifer M. Lee
Student,
USC Genetic Counseling Program,
Columbia, SC

Fundamental Obligations

To the Editor:

I would like to respond to Scott Polzin's comments (*PGC*, 13:4) regarding my thoughts on the issue of prenatal diagnosis solely for the purpose of sex selection (*PGC*,13:3). Mr. Polzin raises a number of interesting points. He seems to believe that if a particular activity is sanctioned by the laws of the land, we have no right to question that behavior. I would remind him that the extermination of Jews, homosexuals and other "undesirables" was the law of the land in Nazi Germany. People of conscience refused to participate in the genocide. In their culture they were punished, often with the loss of their lives, for failing to carry out the law. It seems to be a clear example of a situation where a universal moral code can be easily defined and where the local behavior is morally wrong. I believe this gives credence to my statement that morality is not relative to culture.

Mr. Polzin apparently believes that there is no universal moral code. I think that he fails to appreciate that there are numerous examples of cultural behaviors, policies and laws that are just plain wrong. For example, in some segments of Indian society a widow was expected, until modern times, to throw herself on her husband's funeral pyre; Josef Stalin condemned people to the Gulag for disagreeing with official government policy, and millions of Soviet citizens "disappeared;" the Iraquis are systematically exterminating the Kurds; the Soviet government was responsible for the murders of tens of thousands of Afghanistani children; family honor killings, in which a woman who has "shamed" her family (e.g. by conceiving outside of marriage) are everyday occurrences in segments of Arab society. All of these situations are examples of immoral behavior. I would certainly hope that no one needs a book of "Universal Moral Codes" to appreciate this fact.

Mr. Polzin conveys an attitude among some genetic counselors which is particularly distressing. No genetic counselor is obligated to participate in an activity which he or she views as wrong. Many of our members are opposed to prenatal diagnosis solely for the purpose of sex selection, and I would hope that they stand by their convictions and refuse to provide genetic counseling services to such families. There is a significant difference between understanding the practices of another culture and condoning behavior that is wrong.

If abortion on demand is the law, then individuals have the freedom to make decisions that work for them, and no one has the right to deny anyone access. Nevertheless, if we do not personally struggle with these decisions and decide what is right and wrong, then we are failing to meet a fundamental obligation as thinking members of society.

Michael L. Begleiter, M.S.
Childrens Mercy Hospital
Kansas City, MO

• BOOKS •

Fragile X Syndrome: Diagnosis, Treatment, and Research

editors: Randi Jenssen Hagerman, M.D. and Amy Cronister Silverman, M.S.
publisher: Johns Hopkins University Press, Baltimore, 1991, 378 pp.
price: $85.00, hardback.
reviewed by: Allyn McConkie-Rosell, M.S.W.

Fragile X Syndrome: Diagnosis, Treatment, and Research was written to fill a void in the literature for a comprehensive resource on Fragile X Syndrome. The book is organized so many different disciplines can use it as a reference.

The chapter about the clinical presentation of Fragile X syndrome gives a clear picture of both the physical and behavioral phenotypes. For the clinician, there are chapters on pharmacotherapy and a chapter on other X-linked disorders organized in a table with common clinical features, OMIM, and Birth Defects numbers. There is an excellent chapter by Charles Laird discussing his theory of imprinting, and Stephanie Sherman has written a chapter on the epidemiology.

As with most books, information can often be out of date by press time. This is especially true in the rapidly progressing area of DNA analysis. Because the repetitive sequence and methylation studies were published after this book went to press, the chapters on genetic counseling and molecular biology do not include this information. However, both chapters are still worth reading, as the molecular chapter has a concise general discussion of DNA linkage analysis, techniques, and interpretation. The genetic counseling chapter offers a basis for understanding the repetitive sequence and methylation studies and their application to families. It also provides an appreciation for the complexity of the genetic counseling issues and the psychosocial considerations when counseling families in which the mothers may also be affected.

Making the diagnosis is just the beginning of the educational process, not only for families, but for health professionals who are involved with the care and treatment of children and adults with Fragile X. The chapters on interventions with the Fragile X patient are presented so counselors are able to gain an appreciation of the role of other professionals, such as psychologists, educators, occupational therapists and speech therapists. When requests are made for information regarding a specific disorder, counselors are often limited to supplying clinical and medical information. This book provides a reference with specific and practical interventions for professionals.

Since Fragile X syndrome is a common genetic disorder, genetic counselors, regardless of their focus, will have patients for whom Fragile X syndrome is a concern. This book offers a comprehensive source of information and I would highly recommend it. However, the information regarding carrier, prenatal and confirmation studies must be reinterpreted using the new DNA studies.

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A Time to Decide, A Time to Heal

authors: Families who have faced the news of a fetal anomaly with grief and courage
editors: Molly Minnick, M.S.W, Kathleen Delp, A.C.S.W., Mary Ciotti, M.D.
publisher: Pineapple Press, East Lansing, MI, 1991, 72 pp.
price: $4.95
reviewed by: Martha Walker, M.S.

A Time to Decide, A Time to Heal is a booklet designed to help families decide about and cope with terminating a pregnancy in which a genetic abnormality has been detected. The booklet is well organized and covers many relevant topics. Extensive descriptions of grief are appropriately combined with factual information about prenatal diagnosis, hospital procedures and the importance of genetic counseling. Poems and other quotations from parents who have chosen therapeutic abortion are found throughout the booklet.

The need for a second printing of the booklet within one year is evidence of its positive reception. The strong emotional content is appealing to parents having extreme, possibly unfamiliar feelings. Also, the personal contributions help the readers realize that their experience is not isolated. The authors encourage and facilitate patient self-advocacy. Detailed information about abortion procedures enable female readers to anticipate hospital admission, but the euphemistic "interruption of pregnancy" is somewhat confusing. Although the length and lack of illustrations may deter families who are less educated, this is an outstanding resource which medical professionals can confidently give to parents who are making difficult prenatal decisions.

• ORGANIZATION •

Treacher Collins Foundation

This organization of families and professionals provides support and information with a networking list, a newsletter, a booklet, and resource and referral list. Additional information is available from Hope Charkins-Drazin, Treacher Collins Foundation, P. O. Box 683, Norwich VT 05055.

• SUPPORT GROUP •

JOUBERT'S SYNDROME is a genetically transmitted syndrome marked by agenesis of the cerebellar vermis, disturbances in breathing patterns, ataxia, abnormal eye movements and often times mental retardation.

Several mothers of children with Joubert's syndrome have formed an informal support group. Members currently are located in Virginia, Maryland, Minnesota, North Carolina and Michigan.

Families may write: Mary Van Damme, 12348 Summer Meadow Rd, Rock, MI 49880.

• CLASSIFIED • CLASSIFIED • CLASSIFIED • CLASSIFIED • CLASSIFIED •

LOS ANGELES, CA: Immediate opening for BC/BE Genetic Counselor. Faculty appointment.
RESPONSIBILITIES: All aspects of PN, ped, adult genetics. PN: amnio, CVS, MSAFP, teratogens. Ped/Adult: biweekly genetic clinic, monthly NF clinic. Newborn, adult inpt/psych consults. Teaching and pub oppty. Multiethnic, diverse population.
CONTACT: Beth Ann Burt, MS or Lopa Malkan, MS, LAC/USC Medical Center, 1129 N. State St, #1G24, Los Angeles, CA 90033; 213-226-3816. EOE/AA.

OAKLAND, CA: Immediate opening for BC/BE Genetic Counselor with masters in GC, nursing or related field. Experience in developmental disabilities pref.
RESPONSIBILITIES: General GC to develop genetic screening program; coordinate PN test funding program; liaison with local genetic centers; expand genetic services within agency.
CONTACT: Carol Ross, Regional Center of the East Bay (serving SF Bay area), 2201 Broadway, Suite 500, Oakland, CA 94612; 510-451-7232. EOE/AA.

SACRAMENTO, CA: Immediate opening for BC/BE Genetic Counselor. Exp pref. Salary range: $38-52,000, dep on qualifications and experience.
RESPONSIBILITIES: PN counseling and independence in developing research and educ programs; some admin duties.
CONTACT: Douglas Hershey, MD, 5301 F Street, Suite 202, Sacramento, CA 95819; 916-731-4411. EOE/AA.

SACRAMENTO, CA: Immediate openings for 2 BC/BE Clinical Social Workers II (Genetic Counselors). Salary range $2600-3900/mo, dep on experience.
RESPONSIBILITIES: Enjoy professional input into newly reorganized univ PNDx center serving large, multi-ethnic/cultural geographic area; potential to participate on multidisc team in all aspects of PN coun; accessible to univ continuing ed.
CONTACT: Donna Walgenbach or Ann Peterson, UC Davis Medical Center, 1621 Alhambra, Room 2500, Sacramento, CA 95816; 916-734-6124. [Ref # VL2-0178]. EOE/AA.

SACRAMENTO, CA: June opening for BC/BE Genetic Counselor.
RESPONSIBILITIES: All aspects of ped, general coun and case mngmt: CVS, amnio, teratology, MSAFP and hemoglob scrng.
CONTACT: Mark Lipson, MD, Genetics Dept, Kaiser Permanente Medical Center, 2025 Morse Ave, Sacramento, CA 95825; 916-978-1402. EOE/AA.

MIAMI, FL: Immediate opening for BC/BE Genetic Counselor. Experience and bilingual in Spanish/English req.
RESPONSIBILITIES: Clinical GC in large pediatric and PN program as well as teaching, research and community service at major medical-school affiliated teaching hospital.
CONTACT: Laura Powell, Administrator, Dept Pediatrics, Div Genetics, Univ Miami School Medicine, Box 016820, Miami, FL 33101; 305-547-5741. EOE/AA.

CHICAGO, IL: Immediate opening for BC/BE Genetic Counselor.
RESPONSIBILITIES: Work with clinical geneticist/pediatrician in tertiary care hosp with full range of clinical and lab svc; coun and coord pts in general, metabolic, NF and skeletal dysplasia clinics; in-pt consult; supervise GC students; liaison between lab and referring MDs.
CONTACT: Joel Charrow, MD, Head, Sec. Clin Genetics, The Children's Memorial Hospital, 2300 Children's Plaza, Chicago, IL 60614; 312-880-4462. EOE/AA.

CHICAGO, IL: Immediate opening for 2 BC/BE Genetic Counselors. Clinical exp & fluency in Spanish pref, but not req.
RESPONSIBILITIES: Coordinate svc and GC for 1) newly-established Pediatric Ophthalmology, Genetics & Birth Defects Clin at Eye and Ear hosp: provide admin support for ped ophthalmology clinics; work with support groups; participate in clinical research or 2) general PN & ped svc for newly estab Board of Health clinics: focus on services to large, underserved, minority population, family plan and PN clinics; educ clinic staff. Education, outreach & research oppty.
CONTACT: Barbara Burton, MD, Humana Hospital Center for Medical & Reproductive Genetics, 2929 S. Ellis Ave, Chicago, IL; 60616-3390; 312-567-7340. EOE/AA.

KANSAS CITY, KS: Immediate opening for BC/BE Genetic Counselor (50% PN/50% General Genetics).
RESPONSIBILITIES: Rapidly expanding maternal fetal medicine program: CVS, early & routine amnio; U/S; teratology; MSAFP; pregnancy loss; PUBS; fetal transfusion; pediatric and medical genetic clinics; CF, MD, SB, NF specialty clinics; science teacher educ project. Oppty for research & professional devel.
CONTACT: Debra Collins, MS (4023-C) or Lenna Levitch, MS (OB/GYN 3-C), Univ Kansas Medical Center, 3901 Rainbow Blvd, Kansas City, KS 66160; 913-588-6260. EOE/AA.

NEW ORLEANS, LA: Immediate opening for BC/BE Genetic Associate.
RESPONSIBILITIES: Coordinate active program for inpt hospital consultations, outpt clinics, statewide satellite clinic from intake to follow-up to referral; manage PKU families; public & prof educ.
CONTACT: Emmanuel Shapira, MD, PhD, Director, Human Genetics, Tulane Univ School of Medicine, 1430 Tulane Ave, New Orleans, LA 70112; 504-588-5229. EOE/AA.

BOSTON, MA: Immediate opening for Genetics Program Director, MA Dept Public Health. BC/BE, min 3 yrs exp, incl program planning req.
RESPONSIBILITIES: Full professional respon for planning, implementing and coordinating statewide genetics program.
CONTACT: Deborah Allen, DPH, Director Children with Special Health Care Needs, 150 Tremont St, Boston, MA 02111; 617-727-6941. EOE/AA.

WALTHAM, MA: Immediate opening for BC/BE Genetic Counselor. Minimum 3-yr postgrad clinical exp; excellent commun skills; exp with emerging molecular technology a plus.
RESPONSIBILITIES: All aspects of counseling/case mngmt for growing genetic diagnostic service; assist in developing education and marketing programs.
CONTACT: John P. Richard, Collaborative Diagnostics, 204 Second Ave, Waltham, MA 02154; 617-487-7979 x245. EOE/AA.

BALTIMORE, MD: Immediate opening for BC/BE Genetic Counselor. 1 yr exp pref.
RESPONSIBILITIES: Function independently at commun hosp in PN diag prog; assist perinatologist during procedures; coord Tay Sachs Educ/Carrier Test prog; maintain state birth defects reporting prog; liaison with med depts and commun.
CONTACT: Pamela Young, LCSW, Dept Women's and Children's Svc, Sinai Hosp of Baltimore, 2401 W. Belvedere Ave, Baltimore, MD 21215; 410-578-5314. EOE/AA.

COLUMBIA, MO: Immediate opening for BC/BE Genetic Counselor.
RESPONSIBILITIES: Join team in univ setting; all aspects of coun for pediatric, adult, PN genetics; assist in coordinating statewide outreach.
CONTACT: Judith H. Miles, MD, PhD, Univ Missouri at Columbia Hosp & Clinics, 1 Hospital Drive, Medical Genetics, Dept Child Health, Columbia, MO 65212; 314-882-6991. EOE/AA.

GREENVILLE, NC: Immediate opening for BC/BE Genetic Counselor. Faculty position.
RESPONSIBILITIES: Wide range of responsibilities: PN genetic, specialty and participation in satellite clinics.
CONTACT: O.J. Hood, M.D., East Carolina University School of Medicine, Brody

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NATIONAL SOCIETY OF
GENETIC COUNSELORS, INC.
EXECUTIVE OFFICE
233 CANTERBURY DRIVE
WALLINGFORD, PA 19086

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Building, Room 3E140, Greenville, NC 27858-4354; 919-551-2525. EOE/AA.

TRENTON, NJ: June opening for BC/BE Genetic Counselor. Exp in PN, strong pediatric background pref.
RESPONSIBILITIES: PN counseling, incl: CVS, amnio, PUBS, Level II U/S, fetal loss; coord monthly satellite ped clinic w/ clin geneticist. Some teaching.
CONTACT: Carolee Watkins, MS, Coordinator Genetic Services, Mercer Med Ctr, 446 Bellevue Ave, PO Box 1658, Trenton, NJ 08607-1658; 609-394-4026. EOE/AA.

MADISON, WI: June 1 opening for BC/BE Genetic Counselor.
RESPONSIBILITIES: Join large active team in well-estab, comprehensive, multicertified program with primary responsibilities in CVS and biochem genetics program; oppty for flexibility in assignment; oppty for prof educ & training.
CONTACT: Richard M. Pauli, MD, PhD or Catherine Reiser, MS, Wisconsin Clinical Genetics Ctr, 1500 Highland Ave, Madison, WI 53705-9722; 608-262-9722. EOE/AA.

SEATTLE, WA: Immediate opening for BC/BE Genetic Counselor. Masters in GC or related field with min one year exp req (Internship accepted), computer skills desirable.
RESPONSIBILITIES: Coun for triple screen, PUBS, CVS, amnio, congenital malform, preg mngmt, teratogen; facilitate support groups for genetic terminations; oppty for commun/professional educ & research.
CONTACT: Stefanie Uhrich, MS, Program Manager, Dept. OB/GYN, RH-20, Univ Washington, Seattle, WA 98195; 206-543-3767. EOE/AA.

LEGISLATIVE BRIEFS

NSGC SIGNS ON TO AMICUS BRIEF IN SUPREME COURT CASE

The NSGC has joined with a group of professional organizations concerned with reproductive health care to sign on as a "friend of the court" in the case of Planned Parenthood of Southeast Pennsylvania v. Casey. This case, which the U.S. Supreme Court plans to hear on April 22, may clarify the current Court's view on reproductive rights. The Pennsylvania statutes under question in this case define protected life as beginning at fertilization. They also place several obstacles on reproductive freedom, including mandatory waiting periods for pregnancy termination, notification of a woman's husband in most cases and mandatory education regarding fetal development and abortion alternatives. The Court decision is expected in July.

MARCH FOR WOMEN'S LIVES TO BE HELD IN WASHINGTON

There will be a march for reproductive freedom on Sunday, April 5 in Washington, DC. The activities include an interfaith, multicultural worship service at the Washington Monument at 9:30am, assembly at 10:30 at the Ellipse and the March, beginning at noon. Persons interested in organizing an NSGC contingent should contact Trish Magyari.

NSGC CONTINUES TO SUPPORT MEDICAID FAMILY CARE ACT

In the past two weeks, there has been a flurry of activity on S. 1677, the Medicaid Family Care Act. This act would allow states the option to use Medicaid to fund residential alcohol and drug treatment programs for pregnant women and their children. The bill was cosponsored by Moynihan (D-NY) and Bradley (D-NJ) of the Senate Finance Committee. NSGC recently signed on to two support letters to the entire committee, which currently controls this legislation. Senators pivotal to this legislation are: Riegle (D-MI), Rockefeller (D-WV), Breaux (D-LA), Packwood (R-OR), Chaffee (R-RI) and Durenberger (R-MN). NSGC members residing in these states are urged to contact their senators.

Trish Magyari
Legislative Liaison