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CHANGES IN POLICIES FOR SCREENING FOR MATERNAL SERUM ALPHAFETOPROTEIN: IMPLICATION FOR COUNSELORS

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Prenatal detection of open neural tube defects (anencephaly and open spina bifida) has been possible since the early 1970s through the analysis of amniotic fluid alphafetoprotein (AFP). The utility of that procedure as a diagnostic test, however, was limited to women who were categorized as being at high risk for bearing a child with this type of defect, usually because of a positive family history. Although it was an extremely helpful test for high risk women, it did little to approach neural tube defects from a public health standpoint, because fewer than 10% of affected fetuses were carried by women with a positive family history.

The ability to screen for open lesions in a low risk population was made possible by developing a methodology to identify abnormal AFP levels in the blood of pregnant women during the second trimester. Experience in the United Kingdom as well as in this country has shown that the majority of open fetal defects are detectable through maternal serum alphafetoprotein (MSAFP) screening (1-4).

Initially, such screening was restricted to laboratories that agreed to use the testing for investigational purposes only, or to laboratories that manufactured their own reagents. Those restrictions were pursuant to an FDA policy that prohibited reagent manufacturers from distributing AFP reagents for general use. (The prohibition stemmed from an earlier controversy regarding the use of biochemical tumor markers and was not designed specifically to restrict prenatal testing.)

An increasing number of requests for licenses to distribute these reagents prompted the FDA in 1980 to consider loosening its control of the MSAFP screening process. However, as a result of testimony presented at public hearings, FDA reversed its stance and proposed to license kits under restrictive policies that would have been unprecedented. Under the proposed restrictions, the reagent manufacturer, the laboratory, the primary care physician, and the patient would be saddled with a series of responsibilities that many consider extreme.

More public hearings followed the publication of the guidelines, and the FDA spent many months reviewing the testimony. In June 1983 the FDA announced that it was withdrawing its proposed guidelines. The FDA would be responsible for assuring that certain quality standards would be met, but the application of this testing would be left to the discretion of individual users.

The idea of relatively unrestricted use of commercially available AFP test kits has renewed concerns in the clinical community about the potential misuse of this technology. Although it is likely that the approved kits will perform well, that is, provide a reliable measurement of AFP concentration, both the introduction of testing to the community, and more important, the interpretation of test results, will likely be fragmented and poorly controlled. Some states are considering their own regulation of AFP testing to preempt the confusion and disorganization that many health professionals fear will occur.

Because most objections to unregulated testing concern those aspects of the mechanism that include physician and patient education, communication, and counseling, genetic counselors have an opportunity to be key elements in the integration of serum screening on a routine basis. Involvement can take several forms:

1. States that plan to regulate AFP screening within their boundaries should utilize genetic counselors in the development of regulations. Counselors have contacts with the obstetrical community, laboratories, parent support groups, and other resources that would be useful to those developing regulations.
2. Regulations could require that all abnormal AFP results be coordinated through a genetic counselor. That would help to assure that appropriate follow-up and counseling is provided.
3. Genetic counselors could contract to provide coordinating services for laboratories that will offer screening. The availability of a trained counselor to provide program components would be an advantage in what is sure to be a highly competitive area.
4. Similarly, counselors may contract with reagent manufacturers to provide program elements and materials that would complement a high quality assay. The industry should recognize the advantages inherent in acquiring such services.
5. Counselors in private settings may be able to provide freelance services to several competing laboratories in a defined geographical area that has a high population density.

At the very least, counselors will need to assist in the education of physicians unfamiliar with the AFP screening process. As Holtzman and his colleagues demonstrated (5), physician knowledge about the management of abnormal AFP values may be inadequate. The American College of Obstetricians and Gynecologists, in a news release dated 17 June 1983 (6), was less than enthusiastic about MSAFP screening on a nation-wide basis, but stressed the need for "competent genetic counselors" as part of any legitimate screening effort.

Although it is impossible to predict clearly the direction that MSAFP screening will take, one can reasonably assume that genetic counselors will be essential components of those programs likely to be accepted by the medical community.

1. United Kingdom collaborative study on alphafetoprotein in relation to neural tube defects: Maternal serum alphafetoprotein measurement in antenatal screening for anencephaly and spina bifida in early pregnancy, *Lancet* i:13, 1977.
2. Macri JN: A screening program for neural tube defects in Nassau County: The first year's experience. In *Proceedings of the First Scarborough Conference,* Haddow J and Macri JN, eds., p. 1, 1978.
3. Gardner S, Burton BK, and Johnson AM: Maternal serum alphafetoprotein screening: A report of the Forsyth County project, *Am J Obstet Gynecol* 140:250, 1981.
4. Milunsky A, Alpert E, Neff RK, and Frigoletto FD: Prenatal diagnosis of neural tube defects, IV: Maternal serum alphafetoprotein screening. *Obstet Gynecol* 55:60, 1980.
5. Holtzman NA, et al.: Deficiencies in obstetricians' knowledge of alphafetoprotein screening for neural tube defects. Presented at the annual meeting of the American Society of Human Genetics, Norfolk, VA, November 1983.
6. American College of Obstetricians and Gynecologists: AAP and ACOG statement on alphafetoprotein testing. Washington, DC, 17 June 1983.

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BOOK REVIEWS

Pediatric Research: A Genetic Approach, Edited by Matee Adinolfi, Philip Adinolfi, Philip Benson, Francesco Gianelli, and Mary Seller, London, Spastics International Medical Publications, with J.F. Lippincott Co., Philadelphia, 1982, 245 pages, $27.00.

A Genetic Counseling Casebook, Edited by Eleanor Gordon Applebaum and Stephen K. Firestein, New York, The Free Press, 1983, 291 pages, $19.95.

Whether or not one is preparing to take the examinations for the American Board of Medical Genetics in June 1984, these two publications merit attention from all professionals who provide genetics services. Taken together, these moderately priced volumes provide an admirable review of both the medical and counseling aspects of clinical genetics.

Pediatric Research: A Genetic Approach, a festschrift for Paul Polani, brings together twelve essays that address diverse topics ranging from the epidemiology of congenial defects to a review of past, present, and potential methods to prevent and treat biochemical disorders. The best aspect of the essays is that the individual authors have taken care to provide extensive bibliographies and present sufficiently detailed reviews of the developments in their respective research areas. This format allows the reader to appreciate the problems confronted in each area and to anticipate the rationales that will be necessary for further progress in research. The information is condensed in a very readable form that will prove useful not only for personal review, but also for planning educational programs.

Two essays are illustrative of the general tenor of the book. In his essay on population cytogenetics John L. Hamerton presents a thorough review of the developments that led to the establishment of this research area. In addition to presenting data from several population surveys, the essay discusses problems that have confused the interpretation of such data and the possible ramifications of those problems for future population studies. Hamerton discusses two areas in need of additional research; specific causes of nondisjunction, and the appropriate interpretation of individual differences in the context of population studies.

Mary J. Seller contributes an eloquent discussion of the contribution of genetic and environmental factors to the occurrence of neural tube defects. A number of experiments using curly-tail mice (a breed that has a high genetic susceptibility to neural tube defects) was undertaken to isolate genetic versus environmental contributions to the occurrence of these defects. The role of uterine environment, exposure to a known teratogen, and the interaction between variables are examined in detail. The experimental findings are discussed in light of recent attempts to reduce recurrence of neural tube defects in humans through vitamin therapy.

The second publication, *A Genetic Counseling Casebook,* deals exclusively with counseling issues confronted in clinical genetics. Other publications have outlined counseling techniques, addressed counseling problems associated with specific disease categories, or provided brief excerpts from individual counseling sessions. This volume offers a unique approach in providing detailed accounts of twenty-four counseling interactions that are interspersed with the cogent comments and interpretations of a psychoanalyst. The case reports are written by the involved counselor in all but one case, where the counselee details the narrative.

The in-depth reporting on such a large number of interactions allows the reader to examine varied approaches for addressing many familiar counseling dilemmas. This book does not provide specific recommendations regarding how best to handle a given problem. However, the careful reader will gain general insights by considering how he or she would handle the same problem, how other families have reacted to a similar situation, and how the annotations either agree or disagree with one's interpretation of the narratives.

Aside from providing the reader with the benefits described above, perhaps the most important impact of this publication will be its effect on the form of the genetic counseling literature. There is a very great need for the type of analysis and case narrative presented in this volume. If genetic counselors have as a goal the improved provision of genetic services, they need to examine their counseling experiences and practices in a formal manner and to share those experiences and practices with their colleagues. This useful volume provides an example of one way to accomplish that obligation.

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Philadelphia, PA 19104

NSGC NEWS

GRANT APPLICATIONS APPROVED

The National Foundation for Jewish Genetic Diseases, Inc. (NFJGD), provided grant support for the annual meeting of the Board of Directors of the National Society of Genetic Counselors, Inc. held on 2 November 1983, in Norfolk, Virginia. During that session, the board planned committee projects, regional activities, and national goals for 1983-84. The grant award by the NFJGD represented the third year of financial assistance for the board meeting and was used to defray the costs to board members of an additional day's lodging and meals in Norfolk.

The March of Dimes has approved grant monies for the fourth annual NSGC Education Conference to be held on 15, 16 June 1984 in Denver, Colorado, in conjunction with the Birth Defects Conference. This year's topic will be "Strategies in Genetic Counseling: Issues in Perinatal Care" and will feature invited speakers and member presentations. March of Dimes funding has supported NSGC annual conferences since their inception in 1981 in San Diego.

NSGC MEDIA CENTER

The media center for the next NSGC education program, "Issues in Perinatal Care," will be comprised of books useful to our patients. We plan to compile a list for all age ranges, dealing not only with information (for example, what the patient needs to know about a given diagnosis), but also with the emotional/personal response (for example, *Journey*, by Robert and Suzanne Massie, or *When Bad Things Happen to Good People*, by Harold Kushner). We hope to keep the list manageable by dealing with aspects related to the perinatal period, but do not want to limit it solely to that period, because there are many worthwhile books for patients dealing with birth defects/genetic diagnoses that occur at other times during their lives.

Please send titles of appropriate books to:

Vickie L. Venne
Genetics Service
Children's Hospital
8001 Frost Street
San Diego, CA 92123
(619) 576-5808

Michelle A. Fox
UCLA Medical Center
Division of Medical Genetics
1083 LeConte Avenue
Room MDCC 22-499
Los Angeles, CA 90024
(213) 206-6581

CALL FOR NOMINATIONS

Enclosed in this issue of *Perspectives* is a form for submitting nominations for the NSGC offices of president-elect, secretary, and representatives of regions II, IV, and VI. All members of the society are encouraged to submit names of potential candidates. Your participation in choosing the leaders of the organization is essential to the nomination process.

The members of the 1983-84 nominating committee and their respective regions are: I, Edward Kloza; II, Virgina Corson (chairperson): IV, Elizabeth Thomson; V, Jacqueline Hecht; and VI, Richard Apostol. The deadline for receipt of nominations is 15 February 1984; forms should be returned to Elizabeth Thomson.

Regions for which representatives will be elected are: II (NY, NJ, PA, DE, DC, MD, WV, VA, Quebec, Puerto Rico, Virgin Islands); IV (OH, IN, IL, WI, MI, MN, MO, IA, NE, KS, Ontario); and VI (WA, OR, ID, NV, CA, AZ, HI, AK, British Columbia).

ANNOUNCEMENT OF MEETING AND CALL FOR ABSTRACTS

"Strategies in Genetic Counseling: Genetic Counseling Issues in Perinatal Care," the fourth annual Professional Education Conference of the National Society of Genetic Counselors, will be held 15, 16 June 1984, at the Regency Hotel in Denver, Colorado. The conference will precede the annual Birth Defects Conference, sponsored by the March of Dimes Birth Defects Foundation.

The purpose of the conference is to promote education of and communication among genetic counselors and related professionals. The impact of genetic and congenital problems in the perinatal period—and appropriate intervention strategies—will be addressed through workshops, lectures, and panel discussions.

Abstracts from NSGC members and sponsored non-members will be accepted until 1 March 1984. Papers or data may have been previously presented or published. All abstracts should be sent to Susie Ball, Crippled Children's Services, Yakima Valley Memorial Hospital, 2811 Tieton Drive, Yakima, WA 98902.

Abstract forms will be enclosed with the registration mailing. Individuals who are not on the NSGC mailing list should contact Gayle Mosher, Center for Human Genetics, University of Nebraska Medical Center, 42 & Dewey Avenue, Omaha, NE 68105.

For additional information, contact: Helen Travers, Mailman Center for Child Development (D-820), P.O. Box 106820, Miami, FL 33101.

GENETIC COUNSELOR DESCRIPTION

In response to inquiries from other professionals and students the board of directors has prepared a description of the role of the genetic counselor. This definition will be made available to the executive office of the American Society of Human Genetics, the National Center for Education in Maternal and Child Health, and other interested institutions. In addition, the description will be incorporated into the revised NSGC brochure.

The Genetic Counselor

Genetic counselors are health professionals with advanced education and experience in the areas of medical genetics and counseling. They come from a variety of disciplines including biology, genetics, nursing, psychology, public health, and social work.

Genetic counselors participate in many aspects of health care. Most counselors work as members of a health care team that provides information and support to families who have members with birth defects or genetic disorders and to families who may be at risk for such conditions. They identify families at risk, investigate the problem present in the family, interpret information about the disorders, analyze inheritance patterns and risks of recurrence, and review options available to the family. Genetic counselors also provide supportive counseling to families, serve as patient advocates, and refer families to other support services. They serve as educators and resource people for other health professionals and the general public. Many engage in research activities in the field of medical genetics and genetic counseling.

Health professionals who have had advanced education and experience in the area of medical genetics may be eligible for certification as genetic counselors by the American Board of Medical Genetics. Additional information about the genetic counseling profession may be obtained by contacting the National Society of Genetic Counselors, Inc.

Adopted by the National Society of Genetic Counselors, Inc.
1982-1983 Board of Directors
19 June 1983.

1984 RATES FOR *PERSPECTIVES IN GENETIC COUNSELING*

Beginning with Volume 6, Number 1 (March 1984) of *Perspectives*, the following rates will apply for announcements of positions available, advertisements, subscriptions, and back issues:

Announcements of positions available	$25.00
Advertisements:	
Commercial 1/4 page	40.00
Commercial 1/8 page	25.00
Not-for-profit organizations	at cost
Subscriptions:	
Individuals	15.00
Institutions	20.00
Back Issues:	
per issue	1.25
per volume	5.00

INSTRUCTIONS FOR CONTRIBUTORS

Types of Manuscripts Accepted

Authors may submit articles dealing with the varied professional roles of the genetic counselor, counseling case reports, original research reports, articles addressing topics relevant to the profession of genetic counseling, or letters to the editor that deal with professional issues of the society.

Instructions

All manuscripts must be typed and *double-spaced*. Please submit three copies of each manuscript. The author's name, preferred title, address, and business telephone number must accompany all submissions.

Send all manuscripts to:

Joseph D. McInerney, Editor
Perspectives in Genetic Counseling
BSCS
The Colorado College
Colorado Springs, CO 80903

Deadline for 1984 Issues

March Issue:	1 January 1984
June Issue:	1 April 1984
September Issue:	1 July 1984
December Issue:	1 October 1984

Specific Instructions for Counseling Case Reports

The purpose of counseling case reports is to present organized discussions of the counseling and case management problems confronted in the clinical genetics setting. The format for counseling case reports is as follows:

1. Present a brief statement of the diagnostic information and the reasons for seeking genetic services.
2. Describe the counseling problems or case management difficulties encountered.
3. Discuss how the problems were addressed, including the rationale for your course of action.
4. Present a broader discussion outlining other methods one might use to deal with similar problems.

Sections (3) and (4) should include citations of the counseling and/or genetics literature to substantiate your discussion and methods.

ANNOUNCEMENTS

NORTH JERSEY NEUROFIBROMATOSIS GROUP FORMED

The North Jersey Neurofibromatosis Support Group is proud to announce its incorporation and new chapter status within the National Neurofibromatosis Foundation. Meetings are held on the second Wednesday of each month at the Bergen County Medical Society, Hackensack, New Jersey. For further information please call Donna Oettinger at: (201) 265-2354.

CORRESPONDENCE

To the Editor:

I was somewhat concerned and surprised when I read the article by Niecee Singer ("Influence of Ethnicity, Culture, and Religion on the Genetic Counseling Process," *Perspectives*, September 1983, p.1) which described the content of the regional educational meeting held in New York in May 1983. I found the use of the term "WASP" objectionable. It is not clear to me whether its use was Ms. Singer's or whether she was reporting its use by the speaker whose lecture she was describing.

There apparently are individuals who do not perceive this term as pejorative, but my experience has been that it is frequently employed as a denigrating expression in much the same way as any other ethnic slur. I believe that its use is inappropriate and unprofessional. It seemed to me to be particularly ironic that in an article describing the importance of sensitizing oneself to the differences that may exist between various ethnic and cultural groups, this term was considered acceptable, not only by the author/speaker, but by the editor.

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New York University Medical Center
New York, NY 10016

Niecee Singer replies:

It was evident to the audience at the educational conference that the speaker used the term "*so-called* WASP" (author's emphasis) as a descriptive term, not a disparaging one. The focus of his presentation was the tendency to stereotype individuals according to their ethnic backgrounds. The recognition that a person who is white, Anglo-Saxon and Protestant often comes to the counseling session with different values and goals from a person who is white and Jewish or black and Baptist was an important observation. The speaker applauded those differences and felt they should be cherished, in addition to being understood. If he employed a commonly-used acronym to describe a particular group, it in no way detracted from the excellence of his presentation.

POSITIONS AVAILABLE

Genetic Counselor: To serve as coordinator of the Illinois Statewide Program in Genetics; the individual will be based in Springfield, Illinois. Candidates must be board certified or board eligible. This position is funded by the National Genetic Diseases Act and is available immediately. All inquiries and applications should be addressed to: Celia I. Kay, MD, PhD, President, Genetics Task Force of Illinois, Inc., Lutheran General Hospital, 1775 Dempster, Park Ridge, IL 60068, phone: (312) 696-7705.

Genetic Associate: Position available in medical genetics, University of British Columbia, to commence immediately. The position involves collection of data pre-counseling, counseling of prenatal and specialty clinic cases, the screening of referrals, and follow-up of families. Public education involvement and participation in research projects required. Formal genetic associate training expected (i.e., MS degree), practical experience desired. Send CV and three references to: Dr. J. Hall, Medical Genetics, Grace Hospital, 4490 Oak St., Vancouver, BC, V6H 3V5. The University of British Columbia offers equal opportunity for employment to qualified male and female candidates. Preference will be given to Canadian citizens or Landed Immigrants.

Genetic Associate: The University Hospital of Jacksonville has an opening for a coordinator and counselor for a large prenatal diagnosis component of the genetics division. Candidates must be board certified or board eligible. Experience in amniocentesis counseling is preferred. The position is available immediately; starting salary is $19,000, plus fringes. Contact: Charlotte Lafer, MD, Department of Pediatrics, Division of Genetics, University Hospital of Jacksonville, 655 West Eighth Street, Jacksonville, FL 32209, (904) 350-6872.

Genetics/Reproductive Health Nursing Consultant: The Wisconsin Department of Health and Social Services, Division of Health, has a vacancy for a genetics/reproductive health nursing consultant. Responsibilities include: provision of state-wide nursing consultation in the area of genetic disorders in reproductive health care; development, interpretation, evaluation, and assistance in implementing standards and guidelines for reproductive health care, genetic services, and general maternal and child health programs; in planning, developing, implementing, and evaluating educational programs. Knowledge requirements: principles and practices in maternal and child health nursing, genetics, metabolic disorders, and reproductive health. Special requirements: registration or eligibility for registration as a professional nurse in Wisconsin. Wisconsin residence is not required. Starting salary is between $2,075 and $2,496 per month, depending on training and experience. Send resume to: Joyce Krey, 1 West Wilson Street, Box 7850, Madison, WI 53707, (608) 266-7346.

Genetic Counselor/Associate: The Department of Medical Genetics, Shodair Children's Hospital, Helena, MT, has a position available 1 July 1984. This is a new position in a general clinical genetics program providing state-wide services, including clinic coordination, general genetic counseling, public education, and counseling for prenatal diagnosis, teratology, and fetal death. Candidate must be board eligible/certified by the American Board of Medical Genetics. Send resume to: John M. Opitz, MD, Shodair Children's Hospital, Box 5539, Helena, MT 59604, (406) 442-1980. Shodair Children's Hospital is an Affirmative Action/Equal Opportunity Employer.

Coordinator: The University of British Columbia Department of Medical Genetics and the Huntington's Society of Canada have a position available for a coordinator for their assessment and referral program for Huntington's disease. The successful candidate will be expected to provide and/or arrange counseling and supportive services for involved families from the Province of British Columbia. The candidate will also participate in ongoing clinical research programs. Closing date for applications is 1 March 1984. Qualifications: preferably a Masters or PhD in genetics, social work, psychology, or nursing. Salary is dependent upon experience and qualifications. In accordance with Canadian immigration requirements, priority will be given to Canadian citizens and permanent residents of Canada. Equal opportunity will be given to male and female applicants. Address curriculum vitae to: Dr. Michael Hayden, Department of Medical Genetics, Grace Hospital, 4490 Oak St., Vancouver, BC, V6H 3V5, Canada, (604) 875-2157.

NEW JOBS HOT-LINE NUMBER
Linda Nicholson: (302) 651-4234

Send all mailing address changes to:

Ann C.M. Smith, NSGC Membership List
Genetic Services, The Children's Hospital
1056 East 19th Avenue, Denver, CO 80218.

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