PERSPECTIVES IN GENETIC COUNSELING

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THE ROLE OF A VOLUNTARY HEALTH AGENCY IN PUBLIC AND PROFESSIONAL EDUCATION IN GENETIC DISEASES AND GENETIC SCREENING

Robert J. Beall and Sherry L. Keramidas

During the next decade, technological and scientific advances are likely to result in development of methods for identifying persons heterozygous for cystic fibrosis (CF) as well as other serious genetic conditions. The introduction of these methods will raise several important issues related to genetic screening and genetic counseling. Fortunately, many voluntary health agencies, such as the Cystic Fibrosis Foundation, may be able to utilize their unique resources to ensure that screening programs are implemented effectively. This article focuses on the role the Cystic Fibrosis Foundation has identified for itself in preparation for the introduction of genetic screening.

The Cystic Fibrosis Foundation: Brief Overview

The Cystic Fibrosis Foundation is dedicated to improving the length and quality of life of the CF patient and ultimately to preventing and/or curing the disease. The foundation, through its national headquarters and network of more than 65 chapters across the United States, supports programs in scientific and medical research; patient care, through a national network of 127 clinical centers; and public and professional education.

Although screening for CF is not available, many CF centers already offer genetic counseling services to at-risk families and the general public. At CF centers, genetic counseling often may involve consultation with the health care team, including physicians, nurses, genetic counselors, and social workers. However, experience with other disorders has indicated that until reliable methods for detection and antenatal diagnosis become available, the present counseling system will have little impact on reducing anxieties about conceiving a child affected by CF. While it is not possible to predict when screening methods will be available, the Cystic Fibrosis Foundation already has begun to explore the scientific, educational, ethical, financial, and political aspects of screening.

Evolution of CF Screening Efforts

Efforts to develop screening tests have grown out of nearly two decades of searching for a CF "factor." Because of the generalized exocrine gland dysfunction in CF, scientists postulated that some circulating factor, produced as an abnormal gene product, could be causing the pathological consequences of the disease. Additional support for this theory came in 1967, when investigators detected in the sweat of CF patients a factor that inhibited sodium reabsorption in rat parotid and human sweat glands.[1] Since that initial report many other factors have been postulated and studied. None, however, has been validated in a reliable screening mechanism.

Despite lack of solid success to date, the search for the elusive CF factor has two important aspects that sustain the interest of investigators. First, in looking for a factor, scientists may unlock the mystery of the basic defect in CF, thereby providing new insights into the pathogenesis of the disease and new guides for treatment of the affected individual. Since the goal of the foundation is to find a method for controlling CF, research will continue in this area. Second, pursuit of the CF factor has generated research into screening methods for both carriers and prenatal diagnosis. The level of research has grown rapidly within the last five years, both in the United States and abroad.

During that period, research on CF screening has been marked by promising leads followed by disappointing results. Several potential methods for detecting asymptomatic carriers of the CF gene have been reported in the professional literature.[2-7] One such method was also reported in the popular press and aroused considerable public interest and inquiry.[4] Unfortunately, when these tests were subjected to additional controlled and blind-sample testing, methodological difficulties arose and results were not significantly greater than those expected by chance. In addition, many potential methods are based on very sophisticated technology that requires highly-trained personnel, considerable time to obtain results, and a cost unrealistic for routine, large-scale screening.

A potential method for detecting CF antenatally was reported in 1980. The method was based on any assay for levels of MUGB reactive proteases in amniotic fluid, which appeared to be lower in samples taken from pregnancies that subsequently resulted in an affected child. The investigation utilized the Cystic Fibrosis Foundation center network to collect amniotic fluid samples from nearly 80 high-risk pregnant women (i.e., women who had previously given birth to a child with CF). After more than half the pregnancies were brought to term, it was apparent that this method could not be utilized for antenatal detection of CF, as the number of false positives and false negatives was unacceptably high. The foundation and its medical advisory council monitored the results of this method from the early stages of its development and testing. Physicians and other health care providers received repeated warnings about the experimental nature of the method, and the inability to make family planning decisions based on its results. Tragically, however, a number of individuals suspected of carrying CF affected fetuses elected to terminate their pregnancies. The

subsequently determined unreliability of the prenatal diagnostic method, and the inability to confirm CF in an aborted fetus, suggest that some of those fetuses may not have been affected.

The experience in developing CF screening methods is not atypical in biomedical science, where what appears initially to be promising ends up as a blind alley. However, these apparent setbacks constitute a basis for new research. There is renewed excitement as rapidly emerging technologies—such as gene mapping and splicing, recombinant DNA methods, and monoclonal antibody techniques—are applied to CF research, and as the pool of scientists working on these challenges continues to expand.

The road to development of reliable screening methods may be long and tedious, and that development is only the first step in establishing CF screening programs. The psychosocial, financial, ethical, and legal aspects of screening also must be considered. Finally, we must ensure that a concerned public is not faced again with disillusionment and disappointment caused by a perceived lack of success of the scientific process.

CF Screening Programs—Potential Magnitude

Eventual implementation of screening for CF will probably represent the largest genetic screening program ever established in the United States. That is apparent when one considers that CF is a chronic, degenerative disease that leads to premature death and is a financial and emotional burden affecting families, that there is currently no cure for CF, and that unlike some other genetic disorders there is no ethnic or racial clustering of the gene. While the incidence of CF is highest among Caucasians, there is still a significant incidence among other racial groups. Thus, carrier screening could be utilized by the entire U.S. population of childbearing age. The enormity of a potential CF screening program is easily demonstrated using U.S. population estimates for 1985.[9] Assuming 57 million females of reproductive age (14-44) and a carrier frequency of approximately 1:20, there would be 8500 at-risk pregnancies and 2125 affected fetuses per annum.

The total number of screening tests performed will depend on the availability and cost of the test. It is conceivable that screening could be performed on the entire population under age 44, or on the population of childbearing age. Other schemes might involve initial screening of women of childbearing age, women contemplating pregnancy, or women who become pregnant, followed by screening of spouses if the female is identified as a carrier. Assuming an estimated average cost of $10.00 per test, the cost of screening may run from $35 million annually, if pregnant females and spouses of female carriers are screened, to nearly $1 billion annually, if the entire population of childbearing age is screened.

Since it is possible that an antenatal diagnostic test will be developed in conjunction with carrier detection, the financial aspects of antenatal testing also must be considered. Assuming 8500 at-risk pregnancies per annum (i.e., both parents are CF heterozygotes) and a minimal cost of $300 per antenatal test, this aspect of screening would cost more than $2.5 million per annum.

The potential scope of CF screening raises several issues, including the need for screening that is accessible to the general public; the need for facilities equipped to handle the potential sample load, yet provide accurate results; the need for a support network to disseminate information about screening and provide counseling services before and after screening; and the need to ensure that the economic incentives of screening do not create entrepreneurship that would affect the quality of screening procedures or result in premature introduction of methods.

The Role of the Cystic Fibrosis Foundation Present and Future

The potential benefits of developing genetic screening methods for CF and other inherited disorders are generally well recognized. By identifying carriers of genetic disorders, couples can be counseled about risks of childbearing and receive information about what to expect if they have an affected child. They can make informed decisions about family planning. Additionally, many normal pregnancies might occur that might otherwise be prevented because of fear of producing an affected child. However, as the experience in carrier screening for the sickle cell trait demonstrated, lack of proper information dissemination about screening can result in unnecessary problems, stereotyping, and discrimination. Therefore, voluntary health agencies focusing on genetic disorders must be prepared to grapple with scientific, psychosocial, ethical, and legal ramifications of screening.

In view of its objectives, the Cystic Fibrosis Foundation has already established a multifaceted role in genetic screening, including:

- monitoring the results of basic research that may lead to development of screening methods.
- supporting grants for research on the basic defect in CF, as well as projects that are attempting to confirm previous research results and determine if those findings constitute a basis for screening.
- fostering dissemination and exchange of scientific information among scientists working in this area and clinicians who work with CF families.
- responding to inquiries about the status of screening from the general public, the media, federal agencies, and private firms that may eventually be involved in conducting screening programs or marketing associated products.
- providing educational materials for the general public so that they are aware of the nature of genetic disorders and screening for those conditions. In that capacity, the foundation has helped support development of a genetics curriculum* for the secondary school level, in which CF is utilized as an example of a genetic disorder. Such mechanisms may help ensure that the public can eventually make informed decisions about screening.
- identifying the psychosocial, ethical, and legal ramifications of screening and determining how those issues can best be addressed.

The foundation continues to pay close attention to the various ramifications of screening. The scientific basis for screening for genetic disorders often appears to be growing faster than society's ability to deal effectively with the ethical, psychosocial, and legal issues that surround this area. Mechanisms for dealing with those issues cannot be established overnight and, in fact, often require up to several years of development and implementation.

*The curriculum was developed by the Biological Sciences Curriculum Study in Colorado Springs, Colorado, and will be released by the Cystic Fibrosis Foundation in late 1982.

To begin developing a plan for screening, the foundation established an expert advisory group that has already identified four general areas of concentration. They are:

- legislation and regulation, including preparation of model legislation for consideration at the federal and state levels. Included are issues such as availability of screening services, other services to be provided, protection of individuals from discrimination based on test results, and quality control of laboratory procedures.
- education, encompassing development of materials for federal and state legislators and agency staff, and for medical and lay communities. Materials will address the nature of CF, screening procedures, results and their meaning for the individual, and other services (i.e., counseling) that are available.
- genetic counseling, focusing on development of approaches that will ensure that information is presented in an accurate, understandable format that encompasses the nature of genetic disorders, CF test results, and implications for childbearing. This area also encompasses the need for follow-up procedures.

In the coming months, the foundation will develop and pursue specific activities that address the needs encompassed in the four areas described above. Those activities must be put in place and tested in advance of the introduction of wide-scale screening. The foundation does not intend to market, oversee, or provide screening services. The foundation views those activities as a function of state, local, or private health care delivery systems. However, the foundation does hope to ensure that all issues related to screening are met effectively. This means that the foundation will work to ensure that tests are available for wide-scale use and are accessible; that tests are performed with appropriate, standardized medical procedures; that results are accurate; that participants are counseled properly concerning the results of the test and all alternatives; and that the public is protected from premature introduction of methods that have not been adequately tested and validated.

Implementation of genetic screening programs cannot be undertaken by the Cystic Fibrosis Foundation or any other single group. Effective programs will require cooperation among voluntary health agencies, health care providers, educators,and federal, state, and local health agencies. These groups must work together, share resources, and strive to attain mutual goals.

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The following section of *Perspectives in Genetic Counseling* includes proceedings of the second National Education Meeting sponsored by NSGC, and held in Brimingham, Alabama in June, 1982.

INVITED SPEAKERS

The Challenge of Health Care in the 1980's

Audrey F. Manley
Chief, Genetic Diseases Services Branch
Office of Maternal and Child Health
Bureau of Community Health Services
Health Services Administration
Rockville, Maryland

Reported by Karen Greendale

Dr. Manley presented an historical perspective of federal legislation concerned with genetic disorders and services in the United States. Among the legislation reviewed were the National Sickle Cell Anemia Control Act (1972), the Genetic Diseases Act (1976), and the Maternal and Child Health Block Grant (1981). Dr. Manley discussed various models for coordination of genetic services developed by different states or regions and funded under the Genetic Diseases Act. Essential components of genetic service programs were outlined and relevant activities of the Centers for Disease Control and the National Clearinghouse for Human Genetic Diseases were highlighted. In addition, Dr. Manley shared some demographic data on the use of prenatal diagnosis, newborn screening, and genetic counseling services as reported by 22 states during 1979 and 1980. The estimated rate of utilization for prenatal diagnostic services by women over 35 was 15.5%; more than 130,000 people were seen for genetic counseling.

The second half of Dr. Manley's talk focused on changes resulting from the Reagan Administration's "new federalism" program. Reductions in funding were reviewed. Competitive grant review is now in progress for programs that have been funded for fewer than four years. Dr. Manley is hopeful that most projects that have been funded for more than four years will be continued with a combination of state funds, possible carry-over federal monies (FY '81), institution of third-party reimbursement for services, and patient fees. She noted that disease prevention is a primary objective of the current administration, and that genetic services should be part of that effort. Dr. Manley concluded with a series of recommendations designed to maintain quality genetic services in a time of increasing consumer demand and decreasing federal support. Among those recommendations was continuing a broad-based approach to genetics rather than an approach oriented toward categorical diseases.

Drugs and Teratogenicity: An Approach for Counseling

Jose Cordero
Center for Disease Control
Atlanta, Georgia
Reported by Jill Hager

Dr. Cordero emphasized that drug exposure in early pregnancy is a common concern. Many couples, he said, seek information beyond that routinely provided by their primary physician. Dr. Cordero included the following basic points for consideration when counseling: the need to determine the patient's specific risk; the need for accurate obstetrical history; the need for a good estimate of the gestational age of fetus at time of exposure; the need to establish the age of the mother. When telephoning the CDC for information about drug exposure, it is helpful to know the generic name of the drug, and the dosage. Standard references for such information include Heinonen, Slone, and Shapiro, *Birth Defects and Drugs in Pregnancy, PSG Publishing Co.*, 1977; and Shepard's *Catalog of Teratogenic Agents*, Johns Hopkins University Press, 1980. Computerized data are also available from CDC. Dr. Cordero discussed interpretation of data, and indicated that case reports are not sufficient, and merely raise additional questions. Interpretation of risks, he said, requires epidemiological research that includes both case-control and cohort studies.

Among the many agents discussed by Dr. Cordero were bendectin, spermicides, alcohol, anti-convulsants, lithium, phenothiazine derivatives, chemotherapy, hyperthermia, methimazole, rubella vaccine, malathione, and agent orange. Teratologists and epidemiologists at CDC are interested in surveying the literature for the most up-to-date information, in conducting research, and in serving the public.

Public Relations Is Everybody's Business

Gloria Sterman Goldstein
Assistant Vice President and Director of University Relations
The University of Alabama, Birmingham
Reported by Carol Struckmeyer

Public relations involves the sensitive management of various situations. That definition applies to the daily responsibilities of a genetic counselor, since counseling activities require an empathetic awareness of public reaction. Although all individuals must accept responsibility for public relations, the challenges for the genetic counselor require especially careful consideration. Ms. Goldstein offered suggestions for using public relations principles in those counseling situations that involve important reproductive decisions.

Ms. Goldstein discussed various approaches for responding to inquiries from the press, using a hypothetical case involving prenatal diagnosis of a cytogenetic abnormality. Ms. Goldstein cautioned the audience to avoid hasty responses that might damage the public's view of genetics. Public relations staff at university and medical centers could, she suggests, assume the responsibility for identifying the specific needs of the press and act as an intermediary in these situations. Specific questions might be identified, discussed with all professionals involved, and answered in writing through a public relations liaison. Other options include referring questions to a superior and preparing written responses. Only those questions asked should be answered, since attempts to introduce new information can be detrimental. Ms. Goldstein discouraged use of the "no comment" response, since it can imply guilt. If misquoted, retractions or clarifications are often ineffective, and therefore are not encouraged. In most cases, the press collects information from a variety of sources, yet the presentation of the information is ultimately the reporter's decision.

Other public relations services available to genetics professionals include consultation on pamphlet publication, arrangement of tours, and coordination of special events.

Shared Caregiving: Emerging Role Prescriptions

Brian Stabler
University of North Carolina, Chapel Hill
Reported by Jodi Rucquoi

Brian Stabler, a clinical psychologist, discussed the role of the genetic counselor in the team approach to caring for families. In recognition of the genetic counselor's need for professional autonomy and his/her sensitivity to the perceptions of the genetic counseling profession by others, he had several suggestions for a positive approach to shared caregiving.

Dr. Stabler reviewed several studies that emphasize the psychosocial aspect of disease management and referred to Maslow's hierarchy of needs as a precursor of change in the biopsychosocial mode. This system considers the individual, his illness, and the environment. Needs assessment is an important tool for Dr. Stabler; he surveys colleagues and plans his strategies for management accordingly. A study of his colleagues led him to suggest continued genetics education for the medical community. Dr. Stabler understands well the genetic counselor's role as a medical specialist who combines an awareness of cellular events with the skill required to help patients make important decisions.

Dr. Stabler urged genetic counselors to assess the needs of the patient and the skills of team members, and to match the two appropriately. It is important, according to Dr. Stabler, to beware of competition because it wastes energy, creates territorial boundaries, and is generally counterproductive. The genetic counselor should avoid large team meetings where there is potential for conflict among many professionals, should work on the system and not on the individuals within it, and attract attention to areas of little conflict.

Genetic counselors can benefit from Dr. Stabler's suggestions concerning integration of the genetics professional into the medical team. Increased public education about genetic counseling can facilitate the inclusion of genetic counseling into multidisciplinary medical practice.

CONFERENCE WORKSHOPS

Private Practice for Genetic Counselors?

Reported by Diane L. Baker

Professionals involved in the delivery of genetic counseling services are exploring creative and nontraditional methods of utilizing their skills. Private practice is one option being considered. Beth-Ann Bloom and Kathleen O'Connor demonstrated, through discussion of their positions as genetic counselors, two aspects of private practice genetic counseling, one existing within the formal medical center setting, and a second physically separated from the traditional medical center.

Ms. O'Connor discussed her work in a private prenatal diagnostic service in Denver, Colorado. The staff in this practice includes an obstetrician, a cytogeneticist, a cytogenetic technician, a genetic counselor, a secretary, and a clerk. The practice has performed over 1000 amniocenteses in the past year and one-half. Events leading to the formation of the private practice included inadequate university support of existing services at the medical center (especially regarding secretarial support), problems scheduling ultrasound, and the inability to return profits from the program to the service for improvements and growth. While the group did not change its basic procedure when it established a private practice, the following improvements

were noted relative to the traditional hospital setting: greatly reduced red tape for patients, improved facilities in both waiting and procedure areas, improved efficiency through ready availability of ultrasound equipment, adequate facilities for counseling, and improved employee salary and health benefits, including the potential for profit sharing.

There were some initial difficulties in establishing this type of practice, particularly in determining fee schedules and in learning approaches to delivery of services as a business. There was some misunderstanding on the part of colleagues at the medical center. The staff has worked together to resolve many of these difficulties. Ms. O'Connor stressed that the practice maintains close professional and personal relationships with the local medical center to ensure cross-referral of patients. This private practice prenatal diagnosis provides a highly professional service that is efficient, economical, and challenging for the staff.

Ms. Bloom has worked for the past three years as a genetic counselor at Gillette Children's Hospital and St. Paul Ramsey Medical Center, St. Paul, Minnesota. She works jointly through the pediatric and pathology departments of those hospitals, with a large rehabilitative pediatric service that does not employ a staff geneticist. The service covers a five state area. There was no ongoing genetic consultation prior to the creation of her position. Ms. Bloom discussed some of the advantages and disadvantages associated with this type of job setting. She does not make diagnoses, but works closely with a certified clinical geneticist from an affiliated university medical center, as well as with specialists from her primary hospitals. It was obvious from her presentation that she has developed a very responsible and professional role with the obstetric, neonatology, pediatric, and family practice staffs. She is extensively involved in training medical students, residents, and staff from those departments. She is also involved in the organization of support groups, both for adolescents and for parents, and does a great deal of counseling with adolescents. Ms. Bloom indicates that her close work with hospital staff has helped establish genetics as a part of primary care. She describes her role as being part of a nontraditional team. It is important, says Ms. Bloom, that a genetic counselor involved in this type of setting—within a medical center, but away from a traditional genetics team—work with a high degree of professional responsibility and commitment.

During the discussion period it was suggested that while these two models represent interesting aspects of the private practice concept, neither one is a model for private practice for general genetic counseling. Further discussion revealed that while other variations of private practice exist, there was no one present who had first-hand experience regarding an existing private practice for pediatric or medical genetics.

Genetic Outreach: A Documented Need for Unsolicited Service

Reported by Pat Nichols

This workshop identified the difficulties of providing unsolicited genetic services to families in need. Difficulties commonly encountered in outreach settings include failure of the family to attend clinic, poor understanding of the purpose or facts of the session, and noncompliance with medical recommendations. Problems in delivery of genetic services are created both by the patient and the medical community.

Barriers to the delivery of genetic services are often created in the medical community because of logistics and attitudes. It can be difficult for an outreach hospital or physician to comply with the detailed handling of specimens or consultation procedures. There may be occasional resistance to referring families to professionals outside the established system. The solution to those problems appears to be education. Educational activities can include lectures and workshop for county medical societies; personal contact with physicians and medical personnel when providing patient follow-up (e.g., phoning in laboratory results and discussing briefly the reasons for the referral); providing physicians with educational packets; and repeating each of those measures in a timely fashion. The goal is to facilitate the operation of the genetics team within the existing medical community.

Barriers to the delivery of genetic services are created by many factors in the patient population, including cultural, social, language, and logistic differences. Attitudes toward medical care vary widely in different cultures, and this has tremendous impact on the acceptance of and compliance with genetic services. Long distances can impede not only patients' compliance and attendance at clinic, but delivery of adequate follow-up. Solutions to all those difficulties again lie in effective education. Care providers must become aware of cultural differences and utilize existing community services to facilitate contacts with families. For example, home visits to rural families and incorporation of public health nurses and social workers into the genetics team can provide valuable opportunities for delivery of services. The genetics team cannot function in an isolated manner, and every effort should be made to have personal contact with other agencies and local health care providers.

There is no single formula for the delivery of unsolicited genetic services, but the best approach is to make every effort to know the population and utilize the existing medical structure.

Management Concepts for Genetic Counselors

Reported by Kurt R. Fenolio

This workshop was organized to help the genetic counselor in his/her administrative capacity. According to workshop leader Luba Djurdjinovic, the successful manager must develop a concept of the service as a whole. The administrator must be aware of the hierarchy within the unit. The roles of the various members should be viewed in terms of feedback and interaction. Ms. Djurdjinovic used the analogy of a business setting to organize the discussion of genetic counselors as managers. A business establishes objectives, and managers coordinate staff activities designed to accomplish the objectives. The manager, of course, is ultimately responsible for achieving stated objectives.

Management has the additional responsibility of providing staff members with the necessary information and motivation to accomplish given tasks. There must be continuous evaluation of performance and procedures. Tasks that are not satisfying the objectives must be changed. Good management requires evaluation skills, the ability to make effective decisions, and the ability to judge people. Ms. Djurdjinovic pointed out that effective decision making includes determining how other members of the organization view the decision. In any decision one must be willing to take risks and must recognize the need for ongoing evaluation.

The workshop also helped the participant assess his/her effectiveness as a leader. Participants were helped to identify whether their management style was task-oriented or concerned primarily with manager-staff relationships. Ms. Djurdjinovic stressed that managers must be ready to shift the style of leadership, at times concentrating on relationships while in other situations concentrating primarily on tasks.

Successes and Pitfalls in Organizing Parent Groups

Reported by Beth Fine

This workshop, moderated by Kathy Delp, addressed the experience of different professionals in working with parent support groups. John Carey described his experience with the Support Organization for Trisomy 18/13, a support group based in Utah. The group has a newsletter that is distributed nationally. This group provides support as well as practical information to parents of children with trisomy 18 or 13. These parents are often isolated, since these are rare conditions. This group has managed to provide support to many individuals by telephone, in person, and through the mail.

Andrea Gainey, University of Connecticut, discussed her success in organizing and working with a PKU parent group. This group worked successfully to pass a state law permitting PKU formula to be dispensed by prescription only. As a result, medical insurance reimburses for the formula. Andrea was not as successful in organizing a parent support group for parents of children with Hunter syndrome. She attributes this to the fact that parents of children with this degenerative condition did not share her perception of the need for such a group. She urges counselors interested in beginning support groups to assess the needs and desires of the parents involved.

Prospects for Professional Growth

Reported by Elizabeth Balkite

The role of the genetic counselor has been well defined by the National Society of Genetic Counselors and the American Board of Medical Genetics. However, opportunities for professional growth and development have not been given comparable consideration.

The goal of this workshop was to identify the need for and methods of achieving professional advancement. Participants also attempted to define appropriate professional goals and methods for achieving them. Michael Begleiter presented data from the NSGC Professional Status Survey regarding opportunities for advancement and actual professional achievement. Each panelist presented his or her point of view of prospects for professional growth.

Diane Baker defined professional growth as expansion of duties and responsibilities, combined with economic expansion and growth. Genetic counselors, she said, need to identify those skills that make them unique, such as a general knowledge of genetics, counseling skills, educational skills, and knowledge of the mechanisms of health care delivery.

Sherry Wallace, a former genetic counselor, discussed professional status and potential. She also reviewed the factors that led her to leave the profession. She cited a lack of opportunity for academic advancement; the rigid structure of academic medicine, she asserted, does not allow a person with a master's degree to go beyond a certain level. The level of responsibility, according to Ms. Wallace, was not commensurate with the level of authority. She felt so overextended that she could not do any job well. There was pressure to do research and an emphasis on quantity for grant statistics rather than on the quality of the work. Finally, there was a lack of financial reward. While she did not have unrealistic expectations, her salary increases did not keep pace with the rate of inflation. She felt these issues require resolution if the field is to grow.

Ethical Issues in Genetic Counseling: A Panel Discussion

Reported by Joanne R. Florio

Prior to the conference, NSGC members submitted cases that raised ethical and/or legal questions. Six cases were chosen and presented by the genetic counselor involved in the case. The following panelists then commented on each case: Robert Baumiller; Sara Finley; and John Fletcher. The audience also contributed to the discussion.

The major issues discussed were:

- the individual's right to privacy with regard to receiving additional genetic counseling when new information becomes available.
- whether amniocentesis should be refused if a couple refuses ultrasound;
- the genetic counselor's role and obligation during a family dispute over whether to have amniocentesis;
- whether a genetic counselor is obligated to report suspected child abuse in a case of counseling for sudden infant death syndrome;
- the genetic counselor's responsibility and obligation to the extended family of a balanced translocation carrier when he or she refuses to inform the family; and
- whether the genetic counselor is obligated to disclose to a family information concerning an error made by a physician or laboratory.

As in any discussion of ethical and legal issues, there were a variety of opinions, and more questions were raised than answers given. However, the discussion did allow participants an opportunity to share similar experiences and obtain new insights into approaching these situations.

All agreed with Dr. Finley's opinion that the "genetic" in genetic counseling is the easiest part to handle. Dealing with the patient's anxiety as well as his personal and family conflicts are the most difficult aspects of counseling. The counselor cannot give the patient the full benefit of his or her services if the counselor does not understand those aspects of counseling.

Involvement in Specialty Clinics

Reported by Gayle A. Mosher

This workshop focused on maximizing the genetic counselor's role as a member of a specialty clinic team. Several genetic counselors working in a variety of specialty clinics shared their experiences and agreed that since genetic counselors now provide an accepted medical service, a logical consequence is involvement in multidisciplinary and interdisciplinary medical clinics.

Cynthia Powell defined the multidisciplinary team approach as one that involves family assessment and management by individual professionals, with little or no interaction between team members. There is no integrated planning. In contrast, an interdisciplinary team approach, where team members share information and work as a group to assess and manage families, is an optimal setting for a specialty clinic. There are, however, inherent difficulties; the wide variety of different specialties, personalities, and medical approaches can interfere with effective operation of the clinic.

The panel, which also included Lisa Glinski and Gretchen Landenburger, pointed out that there must be a clinic team leader to facilitate proper operation. Genetic counselors' broad perspective of the family, according to the panel, makes them a logical choice for team leaders. The panel members who are team leaders viewed their role as appropriate and integral to their teams.

Panel members represented a wide variety of clinics, including muscular dystrophy, craniofacial genetics, spina bifida, hemophilia, cystic fibrosis, and osteogenesis imperfecta. One genetic counselor participates in an adolescent medical clinic. Members expressed the need for increased involvement of genetic counselors in specialty clinics, thereby benefiting the genetic counseling profession, the public, and the medical community.

ABSTRACTS OF CONTRIBUTED PAPERS

Field Testing of a Genetic Screening Questionnaire in Family Planning Programs

A reliable genetic screening questionnaire was developed to identify family planning clients with a possible risk of transmitting a genetic disorder or of having a child with a birth defect. The Genetic Risk Scoring Instrument (GRSI) is a two-part, self-administered questionnaire. The first part (phase I) quickly screens all family planning clients. Phase I is recommended for use in conjunction with a standard medical history form. The second part (phase II) offers clients who have identified a genetic concern or who may be at risk for a genetic disorder or birth defect an opportunity to evaluate the concern and/or risk. Phase II is reviewed by the genetic service nearest the family planning service. The genetic service determines whether to offer the client a genetic counseling appointment. In developing a screening tool for use in family planning, it was essential that the questionnaire account for the variety of medical backgrounds of family planning personnel and clinic routines. Therefore, an option grid was provided to direct the family planning health professional in offering the client appropriate information, referral, or recommendation.

The GRSI was pretested in the spring of 1980 in an upstate New York family planning program. During the program year of 1980-81, the GRSI was implemented and evaluated in three family planning programs in New York, Wisconsin, and Texas. During that period, 1311 family planning clients were screened using the GRSI. Of this sample, 37.8% identified a need for genetic information and/or referral via phase II. A reliability study was undertaken at one of the three sites. Seventy-five percent of the sample reported no new information after an interview with a genetic counselor. Of those clients identified by phase I, 34% accepted phase II as the first step to evaluating their concern or risk. Of this group, 41% returned phase II and 42% were seen by a genetic clinic. In addition, the GRSI was shown not only to be effective in screening, but also to expand the base of family medical history by 33.6%. Finally, use of the GRSI in communities such as the Wisconsin site proved effective in determing the prevalence of genetic disease in that community. The GRSI is an effective genetic screening tool when used to encourage early consideration of possible risk.

Luba Djurdjinovic
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Carrier Detection for DuChenne Muscular Dystrophy: An Updated Survey of Current Practice

Duchenne muscular dystrophy is an X-linked recessive condition whose primary defect is unknown. Genetic counseling depends on accurate carrier detection. That is difficult because of the lack of a specific, unequivocal carrier test, and because of variability of expression of serum CK levels. Emery[1] has provided an extensive summary of various methods for improving the accuracy of carrier detection. Many of those methods involve refining the CK curves by sex and age for normals and carriers. Other examples include detection of differences in other enzymes such as PK and LDH, pathological findings in skeletal muscle by light microscopy, histochemistry and electron microscopy, lymphocyte capping, and erythrocyte membrane characteristics. Ionasescu[4] recommends discriminant analysis of ribosomal protein synthesis in carrier detection. Most recently, Monckton[5] et al, have described a method for carrier detection using autoradiography to identify increased uptake in tritiated leucine in some individual muscle fibers obtained by muscle biopsy. While most of these techniques are potentially useful for carrier detection, many may not be suitable for widespread clinical use. The primary dilemma for genetic counselors then becomes one of establishing a protocol that has a balance between feasibility and accuracy. Recent telephone inquiries directed to various muscular dystrophy clinics around the country and to the national office of the Muscular Dystrophy Association (MDA) indicated that there is no such standard protocol. The present study was designed to ascertain which techniques are in regular clinical use. A simple questionnaire has been mailed to 220 clinics listed in the MDA annual report of 1981. Issues covered in the survey include recommended protocol, recent changes in procedures, specification of relatives screened and at what ages, factors affecting completeness of screening, and delivery of genetic counseling. It is hoped that there will be a return rate sufficient to provide a clear indication of trends in current practice and of factors affecting variability among centers.

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2. Lane RJM, et al *Clin chim Acta* 113:75-86, 1981.
3. Smith I, et al *Clin chim Acta* 98:207-316, 1979.
4. Ionasescu V, et al *Am J Med Genet* 5;5-12, 1980.
5. Monckton G, et al *Am J Hum Genet* 33:978-986, 1981.

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Coping With a Genetic Crisis: Activating Social Networks

The focus of this paper is the application of the crisis intervention model to genetic counseling, with special emphasis on the use of professional and natural helping networks. For many families, genetic counseling occurs during a time of crisis. Crisis occurs when a person faces an obstacle to important life goals—an obstacle that seems insurmountable through the use of customary problem-solving methods. A period of disorganization follows a time of upset, with many unsuccessful attempts at solution. Crisis is characteristically self-limiting and constitutes a transitional period that represents both the danger of increased psychological vulnerability and an opportunity for personal growth through problem resolution.

Crisis is triggered by the stress some families experience when they come for genetic counseling following the identification of a genetic disorder in one or more of their children or the occurrence of genetic-related, failed pregnancy. These families may likely be struggling with the loss of pregnancy or the loss that accompanies producing a child that does not fulfill the dream of a perfect baby. Others seek genetic counseling following conception because they have been identified as being at increased risk. For the second group, decisions must be made in a very brief period, with counseling, prenatal diagnosis, and decisions about pregnancy termination or continuation occurring in a period of from five to six weeks. Concomitantly, these families experience the usual heightened developmental processes that occur during pregnancy, as well as accommodation to the likelihood of an additional family member. Various studies have suggested that social support mediates the impact of stress by strengthening the individual's coping efforts. Professional and natural helping networks are useful to the person or family in crisis.

This paper will emphasize the important role for the counselor in helping families use the social supports available as they cope with stress related to genetic disorder.

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Genetic Counseling in the Lay Literature

Public interest in genetics, genetic counseling, and hereditary syndromes is reflected in an abundance of articles recently published in the lay literature. Since it is valuable for the genetic counselor to have an awareness of the level of knowledge of the individual requesting counseling, this paper reviews many articles appearing in magazines such as *Family Circle* and *Readers Digest* during the last three years. The paper alerts the genetic counselor to inadequate information and misconceptions evident in articles written by professional journalists. The paper also reviews many well-written, easy-to-understand articles that might prove helpful to clients.

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Knowledge and Attitudes of Pediatric Nurse Practitioners Concerning Genetic Disorders

This paper summarizes a survey of pediatric nurse practitioners (PNP) regarding knowledge of genetic diseases and attitudes about genetic counseling as a component of health care. A mailed questionnarie was used to obtain data from 1096 PNPs. The majority of respondents demonstrated moderate to poor knowledge of specific teratogens and genetic disorders. PNPs strongly endorsed the inclusion of genetic counseling in primary health care and regarded themselves as ideal providers of supportive care. Implications for genetic counselors are discussed.

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The Prenatal Diagnosis Laboratory of New York City: The First 1000 Counselees

This paper describes the outcome of counseling with reference to amniocentesis for the first 1000 pregnant women seen by the genetic counselors of the Prenatal Diagnosis Laboratory (PDL). These 1000 counselees comprised approximately one-third of the caseload of the PDL in the first two and one-half years of its operation. The PDL is the largest cytogenetic screening program in the United States. In addition to its laboratory facilities, it offers health education services to consumers and health professionals, prenatal diagnostic counseling, and follow-up medical genetics counseling in the event of abnormal test results. The PDL has contracts with 22 hospitals in New York City. PDL counselors have established genetic counseling programs in seven hospitals, where they do on-site counseling. Prior to establishment of the PDL, amniocentesis was accessible only to the middle and upper classes. Socioeconomic, ethnic, racial, and demographic data are presented to demonstrate the impact of the PDL in making amniocentesis available to all socioeconomic groups. Data derived from confirmatory studies and follow-up reports have been documented. Reasons for not undergoing amniocentesis were collected and analyzed, and thoughts are offered concerning some surprising data.

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Genetic Contact Program: Involvement of Local Health Professionals in a Statewide Genetic Services Network Project

A comprehensive statewide population and family oriented network of genetic services has been established in Wisconsin. Its objectives are to

- increase awareness among the professional and lay public of the needs for and benefits provided by clinical genetics
- identify families and populations in need
- document the incidence of birth defects and other genetic disorders,
- organize and provide services through a network of satellite and outreach clinics.

The local base for the network is a unique, statewide organization of community health providers (both physician and nonphysician professionals), representatives from agencies involved with families with genetically determined disorders (Association for Retarded Citizens, Muscular Dystrophy Association, Committee to Combat Huntington's Disease, March of Dimes), clergy, social workers, and teachers. Through this network, new professionals, called "genetic contacts," participate in annual workshops where they receive guidelines for collaboration with their own community as well as with the nearest genetics associate or genetics center. They are also able to identify, communicate with, and refer families to appropriate agencies. A similar contact program for the clergy is being developed. The strength of the contact network in one area of the state has evolved into a regional genetics task force that serves as a local advocacy group for clinical genetics and will also assess and document community need for education and service.

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Job Description: Genetic Counselor

Since the role of the genetic counselor is new, many facilities are struggling to develop job descriptions. Several problem areas have been defined. There is a wide range of education and experience of those presently working as genetic counselors (including genetic associate, social worker, and registered nurse with or without master's degree). The generally low salary may reflect the field's predominantly female membership. A collection of current job descriptions raises many issues that concern genetic counselors. The paper discusses helpful ideas in writing creative, nonrestrictive job descriptions.

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