



PERSPECTIVES IN GENETIC COUNSELING

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GENETIC DISEASES SERVICES PROGRAM: CURRENT STATUS

Audrey F. Manley, M.D.

The Genetic Diseases Services Program is authorized under Part A, Title XI of the Public Health Service (PHS) Act, as amended. The primary objective of the program is to deliver genetic disease information, education, testing and counseling services, and provide medical referral for all persons who are suspected of having, or transmitting, a genetic disorder.

The basic implementation strategy is to build a national genetic diseases program, utilizing the resources made available under the present law in combination with existing state and federal health care delivery systems and provider capabilities, including local, public, and private resources and facilities. The program is designed to support and strengthen statewide and regional genetic services delivery systems. The coordination and linkage of genetics programs with existing health care delivery systems—such as the Maternal and Child Health Programs, Crippled Children's Services Programs, Hemophilia Treatment Centers, and Sickle Cell Projects, as well as entitlement programs like Medicaid and Supplemental Security Income Disabled Children's Programs—are basic to the implementation strategy. The development of state and regional laboratory capability is supported under Title V of the Social Security Act as well as under the genetic diseases authority of the PHS Act. Epidemiological assessment and surveillance, laboratory quality control, and education also are integral parts of the program.

A sickle cell screening and education clinic program has been funded since 1972. The genetic disease legislation enacted in 1976 (P.L. 94-278), superseded the sickle cell disease legislation. The original genetics authorization was for three years and was extended for an additional three years to September 30, 1981 under P.L. 95-626.

Funding for FY 1980 was \$11.567 million, which included \$3.5 million for continuing support of the previously funded sickle cell clinics. During FY 1980, the program funded 34 areawide genetic service grants (an increase of 13 over FY 1979) and 11 sickle cell clinic grants, for a total of 45 projects in 37 states, the District of Columbia, and Puerto Rico. Seven sickle cell clinics, previously funded independently, are now merged with and funded through genetic service grants. The areawide service grants are the heart of the program.

Technical laboratory services are provided through intraagency agreements with the Centers for Disease Control (CDC) in Atlanta, Georgia, in the areas of hematology, biochemistry, and cytogenetics. Services include the development of laboratory standards, training manuals, laboratory courses, bench training, proficiency testing, and conducting evaluations of grant project laboratory structures, staffing, and operations. The CDC also provides consultation and technical services on epidemiological problems, as resources permit.

The National Clearinghouse for Human Genetic Diseases has completed its second year of operation and its activities have increased significantly as awareness of its existence has become more widely known. Major documents produced by the Clearinghouse include a catalog of education and information materials (both written and audiovisual), a Directory of Clinical Genetic Service Centers, and a compilation of state laws and regulations on genetic disorders. Other educational activities include support for the development and field testing of a model genetics curriculum for junior high schools which has been developed under the joint efforts of the Colorado Genetics Education, Testing, and Counseling Project, and the Biological Sciences Curriculum Study; support of general genetics education workshops; and a conference on the genetic training and educational needs of nurses and social workers.

A national Genetic Diseases Review and Advisory Committee has been established and held its first meeting on June 15, 1981. The Committee is comprised of 27 members representing a variety of disciplines and national genetic disease organizations.

Since 1976, five annual reports on the implementation of the Genetic Diseases Act have been prepared and submitted to Congress. In preparation for the fifth annual report, 19 of the 21 areawide genetic service projects reported these genetic services: screening of 1,440,752 newborns; counseling for and performing 16,674 antenatal diagnoses; performing 116,512 carrier tests; providing counseling for 60,243 individuals, and providing education and information for more than 54,988 group sessions and individuals.

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FEDERAL BUDGET REDUCTIONS: THE EFFECT ON GENETIC SERVICES PROGRAMS

Edward M. Kloza, M.S.

In May the National Genetics Diseases Act (NGDA) funds were removed from the prevention block grant and transferred to a specific Maternal and Child Health (MCH) block grant by the Senate Finance Committee. This action, if approved by both houses of Congress, is beneficial, since genetic service programs will not be in a position to compete for funds with other, unrelated prevention programs. Several key questions regarding the continuous and adequate funding of genetic service programs await resolution and are discussed here.

The Reagan administration's proposed block grants in health services and prevention were expected to have a serious impact on labor-intensive programs involving genetic counselors. If such block grants were enacted, not only would the level of funding for MCH and NGDA supported programs be reduced by 25%, but the block grant decision would not specifically earmark funds for MCH or genetic services.

Those states that currently have state-mandated genetic programs (California, Iowa, Michigan, Minnesota, New York, and Florida) have mechanisms in place to receive and effectively distribute funds appropriated by block grants. In all other states, the governor or legislature would be responsible for the distribution of those funds. In those states, mandated programs, such as venereal disease or immunization, would be expected to receive priority consideration. The degree of funding that nonmandated genetic programs received would be highly dependent on the success of local lobbying efforts.

One immediate consequence of this proposed action would undoubtedly have been a reduction in the number of positions currently held by genetic counselors. A survey taken of all genetic service and sickle cell center programs funded in FY 1980 by the NGDA revealed that about 88 such positions are funded nationwide, and funds for several more are being requested. The proposed 25% budget reduction will be harmful in itself; coupled with the original block grant concept, it might be devastating. It is encouraging that a number of program directors felt that the genetic counselors currently employed in their programs have proven so valuable that even if the National Genetic Diseases Act did not get re-funded, every attempt would be made to retain them.

In addition to the obvious problem of intrastate competition for funding after implementation of the block grant proposal, at least three other issues have captured the attention of the many opponents of this plan. The first involves loss of visibility. The high profile currently maintained by most statewide genetics programs would have been diminished in the proposed consolidation. This would have been especially damaging since medical genetics and genetic counseling are beginning to receive long-awaited recognition as board-certified specialties.

Also brought under attack is the Reagan administration's contention that block granting will reduce administrative costs, despite a tradition of ineffectiveness on the part of state health departments in administering similar types of programs. In his testimony before the House Budget Committee, Richard Vedra, vice-president of the Coalition for Health Funding, cited the poor record of the Centers for Disease Control's Health Incentive Program, administered by state public health

departments. There is widespread concern that, if the budget proposals are rushed through Congress and the block grants are made to the states this year, the inability of most states to distribute the funds efficiently will result in an administrative nightmare. Projected increased administrative costs, coupled with a delay in apportioning the funds, are expected to result in an effective net loss of an additional 25% of appropriated funds. One proposed solution is to delay the implementation of block grants for at least 12 to 18 months to give state legislatures a chance to develop an effective mechanism for distribution.

Overshadowing these two concerns is the more immediate problem for those consumers who must rely on federally sponsored programs for genetic services. Since there are only about 100 genetics clinics nationwide that exist on nonfederal funds, the number of individuals and families receiving federally subsidized services is quite significant, as illustrated elsewhere in this issue (Manley).

The cuts in funding for genetic services may have surprised those in the genetics community who recall that Health and Human Services (HHS) Secretary Richard Schweiker strongly supported the NGDA when he served on the Senate Labor and Human Resources Committee. That support helped to double the appropriation for genetic services in FY 1980. While funding of genetic services programs is a major concern to the NSGC membership, those programs, of course, are not the only ones affected by the proposed HHS cuts. In addition to the NGDA and MCH funds, 24 other programs face the same fate.

Despite the early opinion of many Capitol Hill observers that the HHS budget cuts were a fait accompli, some hope has arisen for a more sensible resolution:

Reauthorization of the National Genetic Diseases Act. H.R. 2807, submitted by Representative Henry Waxman (D-CA) would extend the Public Health Service Act for an additional four years and include a modest increase in funding. The bill is expected to make its way through the Health and Environment Subcommittee (chaired by Waxman) of the House Energy and Commerce Committee with little problem. Extensive support from several interested health care provider and consumer agencies, including the March of Dimes and Cystic Fibrosis Foundation, has been forthcoming. No companion legislation has yet been introduced in the Senate.

Reorganization of block grants. Another alternative, which has been accomplished in the Senate, is that of restructuring the block grants to combine MCH and genetics appropriations into a separate block. This will eliminate the inevitable intrastate competition with programs dealing with rat control and substance abuse. The American Academy of Pediatrics, the Cystic Fibrosis Foundation, and the March of Dimes are among the 60 agencies that have supported this effort in the interest of uninterrupted delivery of MCH and genetic services.

Local lobbying in state legislatures. In the event that efforts aimed at implementing an alternative to the proposed budget changes fail, it will be necessary to organize providers and consumers of genetic services in statewide legislative lobbying efforts. Failure to demonstrate the rationale for continued funding of genetic services as a first line of defense against disease will likely result in the reduction and possible elimination of genetic services in some states.

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CLINICAL GENETICS PROGRAMS AND SERVICE REIMBURSEMENT

Robert M. Greenstein, M.D.

The development and delivery of comprehensive genetic services are directly related to the availability of adequate funding for the personnel who deliver those services. Historically, clinical genetic services have been delivered by licensed professionals who were reimbursed under third party payment programs via recognized group, individual, or contract arrangements. These individuals are designated by third party payers, as well as by state and federal statutes, as clinical providers and are thus eligible to receive financial reimbursement at a rate usually commensurate with what is common practice for both the specialty and the geographic region. In the field of clinical genetics, the demand for patient services follows the recent explosion of high technological advance and requires a clinical specialist to bridge the gap between basic science concepts and patient care. The number of M.D. providers of these services is limited. At the same time, the number of master's level genetic associates attracted into an exciting, stimulating speciality continues to increase.

The availability of funding for genetic associate positions has come almost exclusively from soft money grants, beginning with the March of Dimes and most recently from the National Genetic Diseases Act. Since genetic associates have not yet achieved national licensure, they have no reimbursement status with third party carriers and are similar to hospital-based or agency-related social workers in that regard. As we proceed through the inflation and budget conscious decade of the '80s, therefore, we are faced with an anomalous situation: The public's demand for clinical genetic services is increasing at the same time that our ability to build a stable financial basis for the delivery of genetic services is in jeopardy.

In 1978, the various genetic services and the state health departments of the New England states (Region 1) joined together to form the New England Regional Genetics Group (NERGG). In addition to goals for quality, regionalization, improved prenatal diagnosis, and regional education, NERGG also established a committee to study third party reimbursement. All of the genetic centers, as well as the various state health departments, recognized that comprehensive reimbursement for the delivery of clinical genetic services would eventually become one of the most important factors for the longevity of these services. It was understood that there are multiple sources of funding for genetic services, which are themselves comprised of both professional fees and laboratory services. Reimbursement for nonphysician or paraprofessional services, such as those provided by genetic associates, appeared to be supported by numerous but vaguely defined sources. The committee also recognized that it was critical for the future integrity of the delivery process that genetic associates be involved in the reimbursement process.

In 1973 an ad hoc committee of the American Society of Human Genetics recommended that the basis for delivery of genetic services be a team model in order to preserve the comprehensive nature of those services. This concept was strongly endorsed by the Third Party Committee as well as by NERGG. The Third Party Reimbursement Committee therefore set about to develop a questionnaire for distribution to all of the genetics centers in New England. The questionnaire was to serve both as a needs assessment of the types of

services presently being delivered throughout New England and as a mechanism to examine the reimbursement procedure itself. This would include an assessment of the numbers and types of professionals delivering genetic services in New England, their funding sources, their billing procedures at the various centers, and the mechanisms used at the centers to establish service charges.

The information obtained from the questionnaire was presented at the NERGG meeting on December 12, 1980 and will be reviewed briefly here. Fifteen of the 21 completed questionnaires were used in the presentation and represented the major clinical services in the six New England states. Region I represents a population of approximately 14 million, and all six states receive funding to varying degrees from the National Genetic Diseases Act. From those genetic centers reporting, personnel breakdown included 17 M.D.s, 7 Ph.D.s, 12 genetic associates, 3 registered nurses, 30 laboratory technologists, 4 social workers, and 9 fellows. Of the funding for M.D. services, 85% was derived from grants, the rest being distributed from direct hospital salaries or from laboratory budgets. This accounting does not include laboratory technologists, whose salary funding represents a crazy quilt of grants, hospital salaries, laboratory budgets, and medical school positions.

The sources of funding for the entire budget of individual centers was also examined. With regard to the amount of the budget that was reimbursable for services rendered, 40 % of the reporting centers derived more than 50 % of their budget from reimbursable services. At the same time, all centers reported increasing utilization of and demand for genetic services, particularly for prenatal diagnosis through amniocentesis, for services in outreach or satellite units, and for the development of new programs for community screening, especially maternal serum alpha-fetoprotein (AFP) screening. It was therefore disturbing to note that the bulk of the newer services was related principally to the efforts of non-M.D. providers, such as genetic associates and social workers. The

EDITOR'S NOTE:

This issue of *Perspectives in Genetic Counseling* addresses the topic of funding for genetic services. In a time when budget cuts are ominously and rapidly approaching, it is wise to consider what sources of funding now exist, how those sources are being affected by the administration's desire to reduce federal spending, and what creative approaches one might explore to ensure continued, adequate funding for genetic services. The events in Washington concerning the National Genetic Diseases Act compel one to consider new strategies to discover alternative sources for funding of genetic services. Potential sources of continuing support, such as third party payers or private industry, must be examined. We hope that the information provided will stimulate discussion at both the regional and national level and will assist those involved in formulating positions and encouraging active participation in this important area.

availability of federal funding has permitted the creation of additional positions for genetic associates to meet expanding service needs. However, concomitant reimbursability for these services has not been forthcoming. The basic fact remains that expanded services are being provided by genetics professionals who cannot claim reimbursability for services, while at the same time the expanded structure of the genetic centers is being built on soft money, i.e., grant funding. This house of cards is particularly vulnerable to the changing winds presently blowing from Washington in the specter of block grants and reduction in service-oriented programs.

What can be done about this transitional period in the development of clinical genetics programs? Certainly it would be helpful to provide the non-M.D. geneticist provider with legitimate access to reimbursability. Individual accreditation and licensure are one obvious pathway. The efforts of the American Board of Medical Genetics appear to favor that view, although preservation of the genetics team rather than individual entrepreneurs is certainly to be desired.

In lieu of any rapid movement in this area for the moment, how might we best nurture and protect the growth of the genetics team and its service capabilities? There are several options through which team services could seek financial support. These would include lump-sum funding by a third party payer, such as by contract with an HMO or a union health plan; reimbursement of services in toto, similar to contract arrangements, in which a total flat fee would entitle the recipient to all of the team services; granting provider status to entire teams, which also includes the non-M.D. genetic associate; and granting provider status to each individual on the team. Medicare plans require provider eligibility. Traditionally this has been the M.D., with some flexibility maintained at the state and local levels for redefinition of the

types of services provided. The recent re-examination by the Health Care Financing Administration of Medicare funding and its relationship to national health insurance leaves negotiations with that agency in limbo. Blue Cross-Blue Shield on the other hand, allows the development of specific contractual agreements for different provider designations. This is certainly attractive, since it allows the genetics team to be so designated; contracts could therefore be arranged for the provision of services by the team. However, credentialing and accreditation of different types of genetics professionals will still be necessary in order to deal with the many different types of funding sources in this country.

Cost effectiveness studies will be necessary to convince third party payers and insurance companies of the desirability of genetic services. The recent attempt of Wright and Elsas (*Am J Med Genet* 6:315-329, 1980) to apply a cost benefit analysis to a genetic disorder is a good example of the subspecialty fulfilling its responsibility in this regard. However, studies of amniocentesis for maternal age and of maternal serum AFP programs will likely become key items in cost effectiveness efforts if meaningful discussions with insurance companies are to occur.

Additional mechanisms might include the promulgation of either state or national legislation that would require insurance group plans to reimburse for both genetic counseling and amniocentesis; such legislation has been enacted in California in recent years. Since there are only a few large insurance companies who underwrite the vast majority of group plans throughout the country, the cost to the individual subscriber or company for including basic genetic services should not be excessive. Genetics professionals may have to become more politically active and visible in this effort. Such involvement would also afford providers of genetic services the opportunity to work more closely with insurance companies in establishing comprehensive and cost effective reimbursement packages. Finally, other sources of continuing funding for genetic services may come from federal and state grants, particularly those that stimulate regionalization and comprehensive programming. We now realize that we cannot count on soft money to build a stable speciality. Perhaps we should look to private industry for additional funding, since the private sector has a significant stake in the cost effectiveness of health care delivery and in shaping insurance packages for their employees.

The information from the NERGG group questionnaire on third party reimbursement has signaled the fact that genetics programs continue to grow at a rapid rate in response to public demands. This growth has been spurred by federal funding, with a major portion of the services now provided by non-M.D. genetic associates. The critical factor here is recognizing that the expanded services provided by genetic associates are supported mainly by grant funds and are therefore vulnerable to changing national priorities. It is necessary for the emerging speciality of clinical genetics to seek funding stability through reliable reimbursement practices. A concerted national effort by genetics groups should proceed as rapidly as possible before changing national priorities result in damaging cut-backs of existing and planned genetic services.

The following bibliography of additional readings on third party reimbursement for genetic services was prepared by Melanie Pepin, M.S.

Bapat V, Seashore M, Greenstein RM: Reimbursement for services and treatment, in Section IV, Connecticut Genetic Services Program (MCH-071001, October, 1978)

Bergler JH: Development of increased financial support for genetic services. Prepared for George Washington University School of Government and Business Administration, *Issues in Health Resources Allocation*, June 6, 1979

Greenstein RM: Approach to third party payers. Prepared for the New England Prenatal Diagnosticians Meeting, April 2, 1979

Jennings CP: Nursing's case for third party reimbursement. *Am J Nursing* 79 (1):110-4. January, 1979

Lawrence D: Physicians assistants and nurse practitioners: their impact on health care access, costs and quality. *Health Med Care Ser Rev* 1 (2):1, 3-12. March-April, 1978 (85 ref)

Melcher GW, Berini Y: Perspectives on insurance coverage for genetic disorders. Prepared for the National Genetics Foundation, October 5, 1978

Walker A: Panel report—Sources of support for genetics associates: Asilomar East, *Perspectives in Genetic Counseling*, 1(3), September, 1979.

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RESOURCES, Beth A. Fine, M.S.

The quality of the "take home lesson" in a genetic counseling session is indicative of the efficacy of genetic services. Too often we rely on the listening skills and memory of our clients. Visual aids and literature can be useful. The following publications may be helpful.

It's Not Too Late for a Baby, Sylvia P. Rubin, 1980. Available from local book stores or from Prentice-Hall, Inc. Englewood Cliffs, NJ 07632, \$6.95.

Sylvia Rubin must be congratulated on her comprehensive book for parents over 35. Her extensive experience in a prenatal diagnosis clinic is apparent. The book opens with statements by several parents concerning their reactions to pregnancy; there follows a section on the psychology of pregnancy. Genetic counseling, ultrasonography, amniocentesis, and fetal and pregnancy complications are also discussed. Chapters concerning drugs and pregnancy, nutrition, fetal monitoring, methods of childbirth, breastfeeding, and comments from obstetricians complete the book.

It's Not Too Late for a Baby is useful for educated people planning a pregnancy; highly educated and motivated clients will benefit most from the book. Most of the case examples are career women who have delayed having families for various reasons. They are elated to be pregnant; few express doubts, impatience, or fears. The volume of information concerning potential problems might frighten some parents unnecessarily, however.

In general, the book is up-to-date and complete, offering a nice review for genetic counselors. Mrs. Rubin has performed a valuable service by discussing these related topics of concern in one volume.

Learning Together: A Guide for Families with Genetic Disorders. DHHS Publication No. (HSA) 80-5131, 1980. Available in single copies or in quantity, from the National Clearinghouse for Human Genetic Diseases, 805 15th Street, NW, Washington, DC 20005, free.

This 24-page booklet is a unique guide for parents interested in receiving many types of services for their handicapped child. The opening discussion addresses parent groups, including objectives, availability, and strategies for organization. Within that framework, educational, health care (including genetic counseling), and advocacy activities are reviewed. A limited list of resources and organizations is also included.

Learning Together would be a good selection for reading material in the waiting area of the genetics clinic, since it introduces families and health professionals not familiar with genetics to the genetic counseling process and related support services. One might also consider using *Learning Together* as a component of in-service training packets.

Autosomal Chromosome Abnormality: A Cause of Birth Defects, Diane Plumridge, M.S.W., 1980. Available from Genetics Clinic, Crippled Children's Division, University of Oregon Health Sciences Center, P.O. Box 574, Portland, OR 97207, \$7.95.

Diane Plumridge has once again shown her skill at making genetic and medical concepts comprehensible to the lay person. This book was a more difficult undertaking than Ms. Plumridge's *Good Things Come in Small Packages*, because it is intended for a broader population. The applicability to individuals, therefore, becomes somewhat limited.

The book covers basic cell biology, chromosome reproduction, and abnormalities. Detailed descriptions of a genetics clinic visit, the lab procedures involved in cytogenetic analysis and karyotyping, cytogenetic research, and prenatal diagnosis are also included. The final chapters consist of reports from parents, photographs, and a section on available resources. Illustrative photographs and diagrams add to the value of the text.

Although this book is written for the lay person, especially parents, it may be more useful for the genetic counselor as a refresher for explaining autosomal chromosome abnormalities. Several sections are exceptional in their approach. "What Makes the Doctor Suspect There is a Chromosome Abnormality?" includes an excellent explanation of dysmorphic features; "What Should We Expect During the Clinic Visit?" points out that IQ testing is often done for the collection of new data as well as for confirmation of a diagnosis; "Under What Circumstances Should the Rest of the Family Be Examined?" answers questions many parents ask. The photographs and drawings are good visual aids for use during the counseling session.

Detailed discussions of cell culturing and banding techniques, structural rearrangements, and research trends may be more than most parents are interested in. However, highly motivated and educated parents might be anxious to read about many of the topics in this book. For general use, however, Xeroxed sections would suffice. This publication is thorough and well written. Its message will reach more people if used by professionals as a counseling guide.

Aim to Fight Low Expectation of Down's Syndrome Children. Edited by J. Lucille Poor, Ph.D., 1978. Available from North Central Publishing Co., 274 Fillmore Avenue East, Saint Paul, MN 55107, \$3.95.

This parent's manual was prepared by professionals involved in the Down's Syndrome Project of Saint Paul. Their successful infant stimulation program undoubtedly reflects this group's view that children with Down syndrome (DS) can and should be raised at home. Genetic counselors should offer this manual to parents who have accepted their child's handicap, since grieving parents who have not attained some measure of acceptance may be frustrated or angered by the optimistic tone of the book.

The manual begins with general questions and answers about DS. A curriculum for infant stimulation follows, including sections on motor, social-emotional, language, and cognitive development, as well as general stimulation activities. A large part of the book deals with smiling, laughing, and attachment behaviors in normal infants and those with DS. Short chapters on health aspects, dental care, genetic counseling, and volunteer and parent comments close the book. Throughout the text, the reader is reminded that children with DS have the same course of development as normal children, but that the pace is slower.

The optimistic tone of this book is encouraging to those who have opted to raise their child at home. It is well-written and offers simple, well-explained stimulative tasks. Genetics is not discussed in depth. After genetic counseling, this manual may be used prior to the onset of or in conjunction with an educational program.

Genetics for the Health Professional. University of Colorado Health Sciences Center Genetics Unit, 1981. Available from UCHSC Genetics Unit B-160, 4200 East Ninth Avenue, Denver, CO 80262, \$3.50.

Genetic counselors planning an in-service program for satellite clinic coordinators or for health professionals not well acquainted with genetics will benefit from this concise, complete, well-organized manual. The table of contents can serve as a workshop agenda. Certain sections make excellent handouts. The Genetics Unit's publication was developed with input from several experienced genetic counselors and graduate students. This handbook can save time and money for genetic counselors involved in public education. This group has taken the first large step towards decreasing duplication of services and materials. Our profession can surely benefit from this type of service.

FEE SCHEDULE SET FOR FIRST BOARD CERTIFICATION

The American Board of Medical Genetics (ABMG) has received a grant from the H. J. Kaiser Family Foundation to assist in the development and implementation of the Certification Examination for people providing medical genetic services. These funds have reduced the cost of Board certification to the individual from that which was originally projected.*

All eligible candidates will take the general exam. Depending upon their credentials, candidates can take one or more of the five subspecialty exams. The \$200 eligibility application fee covers the cost of the general exam. In addition, the following fee schedule was adopted by the ABMG for the five subspecialties:

\$ 50 Genetic Counselor
\$100 Clinical Geneticist
\$100 Ph.D. Medical Geneticist
\$100 Clinical Biochemical Geneticist
\$100 Clinical Cytogeneticist

The first certification examination will be given on December 9-10, 1981 at nine exam sites. These sites were selected on the basis of the regional distribution of applicants. Eligible candidates will soon be receiving their site assignments as well as a brochure with additional information and a description of the exams.

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President Elect, National Society of Genetic Counselors

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