

Public Sector Genetic Services in Florida and Georgia:

Current Status and Potential Issues Raised by the Human Genome Project

by Ralph W. Trottier, Ph.D., J.D. and Lee A. Crandall, Ph.D.

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Introduction

This study, which is part of a broader on-going investigation of the effects of changing science and technology on public sector genetic services, focusses on the organizational structure and function of public sector genetic services delivery programs in Florida and Georgia. Two ELSI-sponsored conferences influenced the development and focus of this investigation. An initial outline of a research agenda was developed by Drs. Lee Crandall and Ralph Trottier following a conference held in Houston, Texas in March 1991 (Rothstein). An address delivered by Dr. Ellen Wright Clayton on issues in newborn screening along with presentations on legal aspects of the Human Genome Project by Alexander M. Capron, Lynn D. Fleisher and Harold S.H. Edgar were of particular salience to initial ideas (Clayton; Capron; Fleisher; Edgar). Informative essays delivered at the "Justice and the Human Genome Project" conference held in Chicago, Illinois in November 1991 added additional insight into principal issues in the delivery of genetic medical services (Murphy and Lappé).

The objectives of our study were to examine and describe organizational structure and function of current public sector genetic services delivery programs in Florida and Georgia. Public sector genetic services are here defined to encompass newborn screening (NBS) and genetic medical outreach services (i.e., legislatively mandated and/or tax-supported genetic services). A broad array of information was gathered with a view toward understanding how the Human Genome Project (HGP) may affect such programs.

The nature of our research was exploratory. The process involved both structure-seeking -- that is looking at arrangements and relationships among the various parts of public sector genetic services systems -- as well as meaning-seeking -- eliciting thoughts, ideas and interpretations from managers and operators of the systems. The method of this investigation involved detailed information collection and analysis on service/construct modalities from operational viewpoints rather than a study of the behavior of the operators or the consequences of their actions on consumers of services. (We did not consider the latter set of issues to be unimportant but rather to constitute a separate and substantial research study to be conducted in the future.)

The methodologic character of qualitative research is described in the medical sociology literature (Pearlin). Through the use of semi-structured interviews of key operators and administrators in the genetic services delivery system, observations of services in action, examination of operational documents, analysis of laws and regulations, and constant attention to maintaining current information on the ELSI investigations of others, we purposefully "cast a

wide net" to collect information and data with which we could go beyond our focal interest to interrelate it to changes occurring on a broader scale.

We believe that our investigational methods thereby allow informational analysis and conclusions to be drawn along the lines of Geertz's "thick description," and note that this approach is as applicable to the issues studied here as it would have been had we focused (as is more typically the case) on the behavior of services delivery personnel or consumers of their services (Geertz).

While conducting our investigation, we continually challenged ourselves and others with critical questions such as: does genetics have a role in public health? and, if so, of what value is it in that context? Is the public sector an appropriate venue for medical genetic services? What evidence suggests that the role of genetics in public health will escalate/diminish? What evidence suggests that the role of public programs in funding genetic medical services will escalate/diminish?

As may be anticipated in any research endeavor, we found that raising hard questions often leads to defensive and/or curtailing responses without much support for their rationale, spurs controversy, and spawns additional questions. In this report we make every effort to present our views and recommendations while noting where the views of others are different.

Attempting to determine how the science and technology spawned by the HGP may alter public sector genetics is considered by some to be too speculative at this time. We counter this view by noting that the foundation of the ELSI program was based on the need to anticipate the consequences of scientific and technological change. Its most basic task is prediction of possible legal, ethical and sociocultural problems before they are fully manifest.

This report is organized into five chapters. The first briefly explores the broad legal societal and clinical context that frame the two state genetic programs (Georgia and Florida) that we studied. The second describes and compares the legislative framework underlying the two programs and offers recommendations for future legislative directions. The third chapter describes and evaluates the systems of public sector genetic services extant in the two states in the early 1990s while chapter four explores the current and likely future interrelationship of these programs with state Early Intervention programs mandated by the federal government. Finally, chapter five explores ethical and legal issues related to public sector genetic services that currently exist or seem likely to emerge as the result of new technologies and changes in the organization of health care in the public and private sectors.

Chapter I -- Background

Forecasting how the HGP may influence public sector genetic services must be premised upon a thorough understanding of how these programs were initiated, the manner in which they were intended to operate, how they have changed over time, and how they actually operate at present. This inquiry constitutes the foundation for suggesting how HGP derived technologies may best be incorporated for the benefit of the public at large and especially for those who depend on public sector financing and delivery systems for preventive services and/or medical care.

Prior to the commencement of the ELSI program, noted experts in the field of clinical genetics were suggesting that each state should provide certain minimal genetic services for affected patients and their families, including newborn screening and access to state-of-the-art diagnostic and treatment services (Panny & Bernhardt, **). Clearly, the HGP will produce a new state-of-the-art in diagnostic technologies. These new technologies, like those developed in the past (e.g., the Guthrie test), will eventually filter into and impact upon genetic services available in the public sector.

Newborn screening (NBS) is perhaps the best known and most studied component of public sector genetic services (cf: Acuff, 19**; Acuff & Faden, 19**; Andrews, 19**; Bowman, 19**; Clayton, 19**; Elsas & Stevens, 19**). The Council of Regional Networks (CORN) for Genetic Services has, within the past 8 years, produced uniform minimal guidelines intended to assure the quality of NBS programs nationwide (Therrell et al, 19**).

Generally, these programs focus on early detection of biochemical disorders that are, to varying degrees, controllable through dietary measures, as well as hemoglobinopathies and a few other genetic conditions (e.g., hypothyroidism and congenital adrenal hyperplasia) for which early treatment or prophylaxis prevents disastrous consequences.

To some extent, genetic counseling also appears in the public sector. However, there are basic tensions between the ideology and role of genetic counseling and traditions of public health. Funding for public health programs is typically fostered and resulting programs are typically perceived in a context of "prevention." Strictly interpreted, prevention applied to genetics may imply eugenics, i.e., preventing births among individuals whose progeny might otherwise be "affected" by genetic disease. Perhaps to avoid this interpretation, public health genetics is explicitly defined as not conforming to the traditional contagion-control model of public health. We note that the transmission of genetic disorders creates no immediate threat to society. However, increasing public responsibility for funding chronic and restorative care lends credence to expressed concerns that public policy may encourage the limitation of reproduction among some affected persons as "cost-effective" and may use public sector genetic services to pursue this goal.

A recent Institute of Medicine (IOM) report urges a very cautious approach to screening and testing of children (Andrews et al., eds., 1994). but also acknowledges that genetic testing seems likely to become an increasingly important aspect of health and social policy as developments are made possible through DNA typing. The IOM committee advocates voluntary testing/screening of newborns citing CORN data (1990) that show that voluntary NBS programs are as successful or more successful (based upon number of newborns screened through parental consent) than mandatory ones. This view echoes an earlier study (Faden et al., 1982).

One authority opines that "[w]e simply do not know whether the decisions that are made about newborn screening in a political/administrative system and the potential ability of state-run programs to ensure more uniform testing and follow-up actually lead to better results for children than would occur were newborn screening simply another aspect of routine medical practice" (Clayton, 1992a, p. *). In a comprehensive article on newborn screening, Clayton concludes that potential adverse consequences outweigh the benefits of mandatory NBS programs (Clayton, 1992b). From her analysis of the historical perspective of NBS programs and the quick infiltration of new technologies into state-sponsored NBS programs, and like the view of the IOM report, Dr. Clayton urges caution in adoption of potential screening tests that may become available in the near future.

The role of genetics in public health is seen in quite a different light by those who view the contagious disease model of health as too limiting in the context of evolving perspectives of the role of genetics in disease causation (Meaney & Chang, 1992; Meaney, 1992). One view sees a shift in the role of public health from one of regulation to one of rendering broadly available medical services (Grad & Feitshans, 19**). The latter report takes into account carefully drawn analogies between HIV/AIDS and public health approaches to genetic disorders. Like the views expressed earlier, Grad and Feitshans support voluntary screening with full informed consent. The latter report acknowledges, however, that there may be reasons for mandatory testing in some cases.

Whether it is more practical (and more beneficial) to have mandatory screening for some conditions, but not others, is unsettled. In the case of NBS, the preservation of autonomy argument breaks down because the affected person (the newborn) does not have the capacity to exercise autonomous choice. The argument then shifts to examining whether surrogate autonomy should prevail over the *parens patriae* power of the state. Issues become more complex as the purpose of screening for identification of trait carriers versus identification only of affected individuals is debated.

The "rational" (medically beneficial) approach to genetics in public health is said to have arisen from the phenylalanine hydroxylase deficiency screening process -- the phenylketonuria (PKU) story -- that became practical around 1962 (Schull & Hanis, 1990). Shortly thereafter, nearly every state passed legislation to require screening of all newborns for PKU. This embrace of what appears to be legislated practice of medicine was not, however, initiated without significant political pressure (Clayton, 1992). Schull and Hanis (1990) state that "[S]peculation on the future of genetics in public health, even in the short run, is an intrepid, if not presumptuous act" (p.**)

The impact of new genetic technologies may not be measurable for some years to come but the discovery and use of molecular diagnostics is predicted to proceed at break-neck speed (Hoffman, 1994). The number of biotechnology companies involved in research in the U.S. exceeded 1,100 and recorded a total revenue of \$5.8 billion in 1991 (Small, 1992). This demonstrates that the commercial climate is ripe to promote rapid marketing of biotechnological products. One of the major goals of the watch-dog of HGP research, ELSI, is to "[D]isseminate policy options regarding genetic testing services with potential widespread use" (Collins & Galas, 1993). The future structure and function of genetic services within the health care system is a major area of interest in the ELSI research portfolio (Juengst, 1994).

Current literature and our own research findings demonstrate that genetics has a well-established role in public health and that this role is likely to escalate as new technologies become both available and cost-effective. The IOM report of the National Academy of Sciences identifies policy research needs in the area of addressing deficiencies in data on genetic services (Andrews et al., 1993). This report goes on to state that the ultimate goal of scientific advances through genetic research is the eventual prevention of genetic disorders and that there is growing pressure to broaden screening programs. One state reports on the benefits of a targeted screening process for fragile X syndrome (targeted at mentally retarded males) for the purpose of "informing" their families (Nolin et al., 1992). It has been argued that new and very accurate DNA-based diagnostics for this common genetic disorder, which allow for rapid identification of children and their carrier mothers, make a compelling argument for a movement to offer testing to every pregnant woman or woman of child-bearing age (Rousseau, 1994).

The mandates of newly implemented early intervention programs (Public Law 99-457) lend further credence to the view of Grad and Feitshans. HGP research is anticipated to play a significant role in identifying children with developmental delays (Van Dyke & Lin-Dyken, 1993; First & Palfrey, 1994). Meanwhile, restructuring of the local public health system has been proposed as a means to allow specialty services to become a part of public health medical services, underlining the burgeoning public health role in the delivery of medical care to special populations (Koplin, 1993).

The importance of screening for single-gene disorders aside, the impact of genetics in public health is expected to become much broader as the genetic components of multifactorial disease processes are identified. A key issue will be the role that genetic tests will play in the definition, diagnosis, and evaluation of common diseases of great importance to the general population -- cancers, cardiovascular diseases and diabetes melitis (Meaney, 1992). Meaney further states: "Of more concern is the lack of guidelines on what constitutes a minimal genetic services program" (Meaney, 1992, p.**). Meaney argues that efforts should be expended to define the role of state health departments in the genetics area, that genetics should be incorporated into the mainstream of public health services and that the common assumption of the infrequent occurrence of genetic conditions compared to "other" diseases should be dashed. It is predicted that a dramatic expansion of the role of genetics in public health is likely to follow the development of presymptomatic predictive tests for conditions such as adult polycystic kidney disease, hemochromatosis, breast and colon cancers, and Alzheimer's disease as well as other mental disorders (Hofman et al., 1993; Gogel, 1993).

Rapid advances in knowledge produced by the HGP suggest that the commonly held view that genetic disorders are rare may soon change to one that recognizes a component of genetic risk or predisposition for many prevalent chronic conditions (Brown, 1992; King, Rotter & Motulsky, 1992). As this shift in thinking occurs, medical diagnoses that explore the genetic foundations of disease processes will involve greater numbers of adult patients. The capability to detect predisposition to disease onset through genetic analysis, prior to manifestation of clinical symptomatology, will place genetics in an ever increasingly higher priority as a public health concern.

The national importance of genetics as a health-promotion priority is reflected in the Public Health Service's commitment to achieve its broad objectives stated in Healthy People 2000. Among others, these objectives include increasing the proportion of primary care providers for the purpose of delivering preconception counseling on the risks of genetic disorders (Healthy People 2000a) and improving state-sponsored newborn screening programs to achieve a 90% success rate in the total numbers of newborns screened and a 95% rate in follow-up for infants testing positive (Healthy People 2000b). These and other objectives in the report relating to the field of genetics have been reviewed by one author who expresses some dismay about less apparent aspects of genetics that may more concretely align the intention of the objectives with the practical aspects of genetic medical services (Crocker, 1992). Hopefully, despite the lack of legislative mandates to pursue the objectives of Healthy People 2000, the language of this document is not merely precatory but truly reflects its empowering character.

Revision of clinical thinking about the role of genetics in medical care is critical to progressive disease prevention (Baird, 1990; Schull & Hanis, 1990). A critical question at this time addresses the extent to which public sector health services are preparing for preventive intervention made possible through presymptomatic diagnosis. Because they are expected to meet critical health needs of the nation, public health services have an incentive to evaluate the integration of genetics into all aspects of care for chronic disease (Meaney, 1992). The range of information derived by the HGP

may be so overwhelming and the number of people with need to know so great that mechanisms other than those in place at present may be required to ensure adequate delivery of genetic information and thereby to allow people to recognize and act on critical life choices (Andrews, 1992). A component of our research on genetic counseling supports that prediction and lends credence to the potential for developing alternative multidisciplinary approaches to training in genetic counseling (James, 1995).

Current standards used to determine application and utilization of medical technology need to be reexamined (Golbus, 1992). Emphasis on immediate cost-benefit may not yield long-term cost-effective or acceptable outcomes in light of sophistication of diagnostic capability anticipated to result from HGP research.

Information and technology derived from HGP research will undoubtedly change not only how we view the role of genetics in disease, but how we approach the use of genetics in diagnosis and prognosis. It is anticipated that we will be able to determine the roles of genes in multifactorial disease-creating processes and consequently to detect which individuals have disease-predisposing alleles from infancy, or prior to birth (Fletcher & Wertz, 1990).

There remain gaps in understanding the importance of the roles of family, relatives and culture in defining and dealing with genetic issues (Davidson, 1991; Hayes, 1992; Rosenberg et al., 1992). Careful consideration must also be paid to multicultural issues and sensitivity to our society's expanding ethnic and cultural diversity, factors that are closely intertwined with both the biological risk of various genetic disease and the cultural meanings attributed to diseases and to reproductive decisions (Duster, 1990; Fisher, 1992).

Chapter II -- Legislative Framework of Genetics

GEORGIA

The Georgia NBS legislation is found in Title 31 - Health, Chapter 12 of the Official Code of Georgia Annotated (OCGA): Control of Hazardous Conditions, Preventable Diseases and Metabolic Disorders. This diverse chapter describes statutory powers and regulations regarding immunizations, quarantine, medical genetic services, occupational health and safety, importation of birds to be kept as pets, abatement of bath house operations and sales of contact lenses. For reasons that are evident, other than its placement in this omnibus legislation, the genetics component of this title begs for modernization. Code sections concerned with genetics have changed little since their enactment in 1978.

Code 31-12-5 sets forth the framework for establishing a state-wide network of medical genetic services. The Georgia Department of Human Resources (DHR) has the responsibility of establishing components of the network to include training of personnel in genetics, conducting research on genetic disorders, assuring quality control of laboratory services and also has the responsibility to provide for genetic counseling.

Section 31-26-6 empowers the DHR to promulgate rules and regulations to establish "a system for prevention of mental retardation resulting from inherited metabolic disorders." Seven conditions (phenylketonuria, galactosemia, tyrosinemia, hemocystinuria, maple syrup urine disease, hypothyroidism, and congenital adrenal hyperplasia) are specifically listed for which all newborns shall be screened. This section goes on to state "and such other inherited metabolic disorders as may be determined in the future to cause mental retardation if undiagnosed and untreated."

It is curious and worthy of note that the legislation places inherited disorders under a general category of "hazardous conditions." Prevention of mental retardation is the strict (narrow) interpretation of the NBS statute. In 1989, an amendment to the statute added congenital adrenal hyperplasia (CAH) to the NBS list. At that time, the literature reports that only Alaska screened for CAH (Stevens et al., 1988). The incidence of CAH in the general population is reported to be 1:(12-15) x 10³, ranking 5th in a list of 9 conditions for which states reported in their NBS programs (Stevens, et al. 1988). The incidence of CAH is highest (1:680) in Yupik Eskimos. Based upon the number of live births in GA, it was estimated that 9 to 10 cases per year would be detected and perhaps more, if CAH were found to have a greater prevalence in the black population. Based upon the most recent report available, 9 cases of CAH were detected in the NBS program in 1993 and a total of 29 cases have been detected since the start of CAH screening on June 1, 1990 (Comprehensive Newborn Metabolic Screening Annual Report 1993, issued 6/20/94, compiled by Ann L. Brown, RN, MPH, Mary J. Kennedy, RN, BS, and Paul M. Fernhoff, MD, Division of Medical Genetics, Department of Pediatrics, Emory University, submitted to Mary Ann Henson, Genetics Program Manager, Children's Health Services Unit, Georgia DHR). The original prediction made by Louis J. Elsas, MD, not only proved to be accurate but

according to the 1993 report (cited above), CAH ranked 2nd among the 7 metabolic disorders in the NBS profile. By 1990, 8 states reported screening for CAH (NBS Report: 1990, CORN, issued February 1992). A recent article by the U.S. Public Health Service reports 13 states screen for CAH (U.S. Public Health Service, 1994). Neither the Georgia nor CORN CAH data are reported by racial categories. According to 1990 CORN data, the incidence of CAH in the Alaskan population was only slightly higher than that reported for the general population. In 5 of the 8 states reporting, the incidence of CAH fell below that of the general population. It appears that the stated purpose of NBS in GA with regard to prevention of mental retardation is not to be strictly interpreted considering that CAH, although causing serious morbidity otherwise, does not result in mental retardation.

OCGA 31-12-7 directs the GA DHR to adopt and promulgate rules and regulations governing tests for PKU, sickle cell anemia (SCA) and sickle cell trait (SCT) indicating that testing should be based upon susceptibility to these conditions. This same code section allows for exceptions to testing where parents object on grounds of religious beliefs. Part (b) of this section imposes an affirmative duty on the examining physician or the DHR to inform parents if their child is so "afflicted" and in cases of SCA or SCT, that "[c]ounseling regarding the nature of the disease, its effects, and its treatment is available without cost from the department and the county board of health or the county department of health."

Exercising its empowerment, the GA DHR configured its plan for NBS (Rules of DHR - Public Health, Chapter 290-5-12 (March 2, 1983)). Part .3 of these regulations -- "Sickle Cell Testing, Amended," targets SC screening to "[Infants with either or both parents of African, Arabian, Greek, Maltese, Portuguese, Puerto Rican, Sardinian, Sicilian, South and Central American, Southern Asian and Spanish origin which is to be determined by information provided on the informed consent form" (emphasis added).

The sickle cell (SC) aspect of the Georgia NBS program has been referred to as a mandatory/voluntary, targeted system. The law mandates testing for all newborns for 7 conditions (OCGA 31-12-6) including PKU, then provides for PKU and SC to be tested in "susceptible" newborns (OCGA 31-12-7). The plan put in place by the DHR targets along lines of defined ethnic/racial groups. No such plan was devised for PKU in spite of the fact that this condition is rare in some of the ethnic groups targeted in the SC scheme (Scott & Cederbaum, 19**). Of the current NBS tests found in the profile of state programs, SC is the only screen that is treated disparately along ethnic/racial lines.

Questions were raised in Georgia as to whether the DHR, by virtue of its statutory authority, could require SC screening in all newborns, whether the attending physician should make the decision as to who is susceptible to SCD or SCT, and whether the determination of who will be tested may be based on race information appearing on the birth certificates of newborns. These questions were brought to the attention of the Georgia Attorney General in a letter from the Commissioner of the DHR. Strictly interpreting the NBS statute, the Attorney General referred to the "mental retardation" prevention purpose of the statute and opined that unless SCD or SCT could be classified as to cause mental retardation, then the statute does not authorize the testing of all newborns for SCD or SCT. The opinion continues to explain that where it is administratively impossible to determine susceptibility, then the DHR could justifiably require universal screening regardless of the expressly stated or apparent racial classification provided in the statutory language. The opinion notes that the Georgia NBS statute did not impose a rigid requirement on the DHR to absolutely test all susceptible newborns as it allows for a "[n]early as possible" standard. In his analysis, the Attorney General noted that the blood sample taken from the newborn (all newborns) would be subjected to other tests mandated by statute or regulation and that the only difference with respect to SC testing is based upon a determination of susceptibility. There would be no further inconvenience to the newborn in terms of additional blood samples required for the SC test. The opinion concludes that the DHR may restrict testing to individuals determined to be susceptible so long as it establishes clearly defined guidelines for making such determination. The attending physician could make the determination of who is to be tested provided the DHR promulgates defined standards by which the physician is guided in determining the susceptibility. Reliance on information appearing on birth certificates could be one factor in making a susceptibility determination, acknowledging limitations based upon the information being supplied by the parent(s). The opinion concludes that the DHR may require SC screening in all newborns or may restrict testing to susceptible persons (Opinion. Att'y Gen. No. 81-40, May 20, 1981).

A question may be raised as to whether restriction to susceptible classes of persons is, in fact, legally tenable in light of medical practice standards. By assuming the responsibility of mandating medical tests, the state has inserted itself

into the realm of medical practice. Courts have determined that practice standards based upon performing tests in individuals thought to be most susceptible to disease conditions fail where (1) the test is simple to perform, (2) the test is well known,(3) *** (4) the test is inexpensive, (5) the test is not harmful to the patient (interpreted by us to mean "not physically harmful" as compared to "informationally harmful"), (6) the test results are definite, and (7) the disease could be arrested by early detection, and the disease effects, if undetected, become irreversible over time (Helling v. Carey, 88 Wash. 2d. 514, 519 P.2d. 981 (1974)).

The Helling case involved finding an ophthalmologist negligent in failing to administer a glaucoma test to a patient in her twenties. At the time of this decision, administering the glaucoma test to patients over 40 years of age was the standard of care by that medical specialty. This case is interesting for its potential analogy to what may become the "standard of care" with respect to genetic testing. Each item except the 7th under the Helling test appears to be applicable to genetic testing as currently carried out by most states' NBS programs. Although SC does not strictly meet the 7th criterion, it cannot be argued that early detection of SC is of no benefit to the individual and the individual's family. We have recently argued that limiting SC screening to susceptible persons is no longer tenable, if in fact it ever was (Phoenix et al, 1996). As of this writing, the GA DHR has neither required universal SC screening in newborns nor has it issued guidelines or objective criteria as to how attending physicians shall determine SC susceptibility.

Some hospitals in Georgia screen all newborns for SC as a routine matter of care. To gain a better understanding of how the system of informing parents operates as a matter of practicality, we examined the process of how NBS is approached at one hospital. We examined two forms that become part of the medical record. One form (designated as "Notification of Metabolic Disease Screening) recited the requirement for testing all newborns for 6 metabolic disorders. CAH was not listed on the form, perhaps because the form was dated prior to the time CAH was added to the scheme (and the form had not been altered to include this condition). The form explains that the conditions listed result in mental retardation and that a blood sample is taken by pricking the heel of the infant. No details are offered as to any of the conditions listed and counseling is not mentioned on the form. This form did not state that the parent(s) may object to testing based on religious grounds. In spite of the fact that the tests are mandatory according to law, there were lines on the form for parent(s) and witness signature and date. Perhaps the signature is interpreted as an acknowledgment of notification but it appears to be similar to obtaining consent. It is unclear whether refusal to sign the information form is construed as objection to testing. Neither the Georgia NBS statute nor the DHR NBS regulations specify the form of objection to screening. The second form we analyzed was entitled "Consent for Metabolic and Sickle Cell Newborns". Its top portion explained the Georgia law on SC screening and listed the ethnic/racial origin criteria that trigger the requirement to screen susceptible individuals. The form then gives the parent the option of consenting to the screen for SC or objecting based upon religious reasons. The bottom portion of this form recites the legal requirement for metabolic disorder screening and indicates that, in the event of an abnormality, the parents will be notified by either their private physician or the county health department. A statement is then included that acknowledges having read the form (or that the form was read to the patient) and having been provided with the opportunity to ask questions. The bottom portion of the form includes the same "consent" or "object" statements as on the top (SC) portion. In essence, both forms appear to be consent documents when, in fact, the law does not provide for testing based upon consent but, instead allows only for objection to testing, and then only on grounds of religious tenets or practices.

We understand that this confusion has led to treating the GA NBS program as a "voluntary" one rather than a mandatory one, resulting in some infants not being tested according to the letter of the law. The two forms are clear indicators that ethnic/racial groups are de facto treated disparately. We assume that mothers identifying their racial origin as something other than the statutory categories are provided with the form that does not mention SC even though, as provided in the NBS statute, the SC test would be performed upon request by the parent, regardless of race or ethnic origin. We have examined the SC screening policies issue in greater detail and incorporate that information by reference as part of this report (Phoenix, et al., 1996, included as Appendix**).

There is an apparent dichotomy of thought by considered authorities on this matter. On the one hand, universal SC screening recommended by the Sickle Cell Disease Guideline Panel of the Agency for Health Care Policy and Research, U.S. Public Health Service, is endorsed by the American Academy of Pediatrics, the American Nurses Association and the National Medical Association; while, on the other hand, The American Academy of Family Physicians, the Canadian Task Force on the Periodic Health Examination and the U.S. Preventive Services Task Force

recommend screening for hemoglobinopathies in high risk ethnic groups (U.S. Public Health Service).

FLORIDA

The Florida NBS legislation is described in Title XXIX Public Health, Chapter 383, Maternity and Infancy Hygiene, FSA 383.14 - Screening for metabolic disorders, other hereditary and congenital disorders, and environmental risk factors. In contrast to the Georgia statute, the Florida statute is written in a current-day context and appears to allow for greater and more clearly defined flexibility. The stated purpose of the statute is to: "[p]romote the screening of all infants born in Florida for phenylketonuria and other metabolic, hereditary, and congenital disorders known to result in significant impairment of health or intellect, as screening programs accepted by current medical practice become available and practical in the judgement of the department" (FSA383.14(1)). The Florida statute describes mechanisms through which the Department of Health and Rehabilitative Services (DHRS) shall establish a multi-level screening process to assess pre- and post-natal environmental risk factors. FSA 383.14(1)(b) provides for privacy safeguards for procedures and information relative to the operations of the DHRS responsibilities under this code section as established under Florida Chapter 411. "Handicap or High-Risk Condition Prevention and Early Childhood Assistance" and Public Law No. 99-457 (early intervention). FSA 383.14(5) establishes a 12 member Genetics and Infant Screening Advisory Council appointed by the secretary of DHRS. Composition of this Council is specified as:

- 2 consumer members
- 3 pediatricians (at least one of which must be a pediatric hematologist).
- 1 representative from each of the 4 medical schools in the state.
- the Deputy Secretary for Health (or designee.)
- 1 representative from the Children's Medical Services Program Office.
- 1 representative from the Development Services Program Office.

The term of office of members is 4 years. The Council serves in an advisory capacity to identify for DHRS those conditions which should be included in the screening and genetics programs, to evaluate laboratory procedures, and for the evaluation of programmatic operations (FSA 383.14(5)(a,b,c)).

COMPARISON OF PROGRAMS

The Florida statute is much broader in scope than the Georgia statute. PKU is the only genetic condition expressly mentioned in the Florida statute. Unlike Florida, Georgia does not currently have an official genetics advisory council (GAC). However, an unofficially functioning GAC was organized by the incumbent Georgia Genetics Program Manager, Mary Ann Henson, MSN, and has functioned within the DHR Children's Services Unit since 1986. A 5-member executive committee of the GAC has presented a proposal to the director of the DHR Division of Public Health to consider empowering the GAC as an official advisory body to DHR. A copy of the proposed GAC by-laws is incorporated as part of this report (Appendix **). Dr. Ralph Trottier, acting as Vice Chair of the GAC, has suggested that the GAC should make every practical effort to ensure cultural diversity among its membership and that at least one member of the GAC should be a medical ethicist. In contrast to the FL statute, there are no specific provisions for the protection of privacy (information control) in the Georgia NBS statute. The privacy protections in the Florida statute specifically making reference to PL 99-457 are discussed in further detail in a following chapter of this report.

LEGAL BASIS FOR GENETIC COUNSELING

Provisions for genetic counseling as well as provisions for follow-up care are expressly stated in the Georgia NBS statute, if operating budgets allow for such. This is carried out in the Georgia program through a DHR contract with Emory University School of Medicine where a certified, masters degree trained, genetics counselor serves as the contact person for positive metabolic screening cases, while genetic counseling for hemoglobinopathy cases is provided by contracts with the Sickle Cell Foundation of Georgia (SC trait counseling), the Sickle Cell Center at Grady Health Systems in Atlanta (MCH block grant), and the Augusta Comprehensive Sickle Cell Center at the Medical College of Georgia. In addition, there are six SC clinics located in municipalities south of Atlanta.

FSA 383.14(3)(f) directs the Florida DHR to promote the availability of genetic counseling for the benefit of parents, siblings and affected infants. This code section states explicitly ties genetic services to the early intervention program as it provides that plans established under this chapter of the code shall be coordinated with provisions found under PL

99-457. FSA 383.14(4) provides for parental objection by written statement presented to the physician or other persons charged with the duty of administering and reporting test and screens under authority of the statute.

RECOMMENDATIONS

Recommendation 1: Based upon our review of the statutes and their implementation in two states we recommend that specific genetic conditions for mandatory or voluntary screening should not be specified in legislation. Instead, we recommend that advisory bodies, acting in an official capacity be assembled for each state or regional cooperative program for the purpose of:

- defining tests to be included in the program, using a deliberative approach involving robust analysis of medical, ethical, legal, social, and economic issues.
- conducting periodic programmatic review and assessment to include consumer satisfaction and trends in medical practice that indicate that changes in the program are warranted.
- evaluating and making recommendations regarding the applicability of new technology impacting upon service delivery.
- coordinating delivery of genetic services with other components of public sector health services.

These advisory/decision-making bodies should be constituted outside of political (legislative or governmental) influence through recommendations made by appropriate public health operatives. We suggest that such advisory bodies be empowered to direct the state's (or region's) department of human resources or public health division as to the specific NBS tests to be included in or deleted from the program. This would avoid the necessity of having to amend legislation, a generally cumbersome and time-consuming process.

Recommendation 2: We recommend elimination of all legislation that either identifies ethnic subgroups for mandatory genetic screening or makes tests available only to certain subgroups. The advent of an increasing number of tests for conditions that vary in prevalence by ethnicity and the increasing genetic diversity of the population make recommendation of appropriate genetic tests on the basis of superficial ethnic/racial identification poor medical practice. The advisory bodies described in Recommendation 1 should recommend which genetic tests should be provided to all newborns at public expense and whether any of these tests should be mandated for all infants. Tests not universally mandated, but judged appropriate for some ethnic/racial subgroups, should be made available at taxpayer expense, but initiated at the request of clients and their physicians. Appropriate utilization of these tests should be encouraged through education and counseling of affected population subgroups.

Chapter III -- Public Sector Genetic Services

This aspect of our study involved in depth fact-finding and analysis of genetic outreach services provided by state-supported programs in the two focus states (Florida and Georgia). This investigation began in July 1992, a time when HGP research had just begun to escalate exponentially. To determine how this enormous scientific endeavor may eventually impact upon public sector genetic services, it seemed logical and prudent to evaluate the structures and functions of these services in their current day operations.

Other than the minimum data sets compiled by CORN (some regions not providing complete information on genetic services), and one published report describing public sector genetic services in the state of Maryland (Panny & Bernhardt, 1989), we have, to date, not identified any other published reports describing comprehensive aspects of public sector genetic services. Although it was stated during the peer-review of our application for continued research that models (other than Maryland) had been clearly documented in the literature (specifically, California, Montana and Iowa), neither a thorough researching of the literature nor direct contact with public sector genetic service operatives in those states revealed any such documentation available or known to the persons who direct and/or operate the programs in those states.

Other views expressed in the same peer-review document opined that our predictions that the public health sector is likely to take on a major role in genetics in the near future are not persuasive, that it is unlikely that the HGP will play a significant role in identifying developmentally delayed children, and that the extent to which public sector health services are preparing for preventive intervention through presymptomatic diagnoses is not currently a critical question. These opinions are directly refuted by our research as well as by current peer-reviewed literature on these

and other critical ELSI issues raised by technological advances rapidly gaining momentum through HGP science.

Initial contacts with key personnel in the Florida and Georgia public sector genetic services programs revealed a cooperative and open invitation to our research endeavor along with expressions of surprise that anyone was interested in such a research enterprise. As noted in Chapter I, our research sought information to address critical ELSI-based questions such as: *Is the public sector an appropriate venue for genetic medical services? How do states' interests (e.g., cost-effective measures for healthy citizenry, prevention of medical expenditures) collide with and produce tension in matters of public policy (e.g., reproductive autonomy). Are there well-known guidelines governing public sector genetic services? If so, what is the evidence that they are employed? Are anticipated outcomes monitored and measured? Is there evidence which suggests that the role of genetics in matters of public health will become increasingly important or escalate with advancing technology in DNA-based diagnostics?*

As we progress in our research, considering these questions and issues expressed or implied by them, we look not only to the findings included at the conclusion of our current grant but also to the continued reports and thoughts published and expressed by others knowledgeable in ELSI research and public sector genetic services.

Throughout the duration of our project period, Georgia did not have permanent leadership in the DHR Division of Public Health or Director of Child Health, issues that drew local press attention ("Leaders Needed for DHR," editorial, *The Atlanta Constitution*, May 17, 1993, A10). Our field research endeavors did not, as a matter of practicality, include views of temporary directorships some of which changed hands during the relatively brief time of conducting our study. The primary emphasis of investigating the Florida/Georgia public sector genetic services systems was to analyze operations from a service construct rather than a study of the behavior of the operators in the systems or the direct outcome or consequences of their actions on the public served.

Our research involved the use of interview and survey techniques plus analysis of systems operations and led to thematic findings which form the basis of ethical, legal and social issue identification. The data collection process employed for the most part in our research was a semi-structured, but unstandardized interview that is commonly used in exploratory studies of this nature (Bauman & Adair, 1992). More highly structured data collection processes were used in aspects of our study soliciting input from district health directors in Georgia. In addition, highly structured questionnaires and interviews were employed the thesis projects conducted by D.C.S. James, Ph.D., S.M. Lybrook, M.S. and K.K. Woodson, M.S., formerly submitted to the D.O.E. E.L.S.I. Division as work products resulting from this grant. Extensive interviews from which key information was obtained were conducted with medical geneticists, genetic counselors, state genetic program directors, genetic outreach nurses, a state district health director, and early intervention personnel. The qualitative data summarized in this report present our analysis of programmatic operations. Quantitative information derived from CORN reports and other sources included to identify specific information (e.g., demographic or indications of system activity) are cited for comparative purposes and were not subjected to statistical evaluation.

GEORGIA

Figure I (following page) illustrates the health district divisions of the 149 counties in Georgia. Tables 1 and 2, following Figure I, describe the population demographics of each health district and health district subunit. The population numbers used in these tables reflect data published in "Georgia Populations Used to Determine Vital Statistics -- 1990" published by the Georgia Vital Records and Health Statistics Office. Each district and district subunit has a health department office. Data in the tables reflect statistics on areas which are not served by organized genetics outreach clinics (Table 1) compared to areas served by the 9 organized public health service genetics outreach clinics (Table 2).

Table 1
Districts Without Genetic Outreach Clinics

DISTRICT	LOCATION	BLACK	WHITE	OTHER	TOTAL
1-1	Rome	29964 (7%)	372741 (92%)	3114 (<1%)	405819
3	Metro Atlanta	679474	1592690	72350	2344514

		(29%)	(68%)	(3%)	
4	Lagrange	95709 (21%)	352671 (78%)	3971 (<1%)	452351
5-1	Dublin	37453 (31%)	82286 (68%)	791 (<1%)	120530
6	Augusta	143855 (38%)	233174 (60%)	7851 (2%)	384880
8-1	Valdosta	58667 (30%)	134322 (69%)	2459 (1%)	195448
1-1, 4, 5-1, 8-1	Excluding Metro Atlanta, Augusta	221793 (19%)	942020 (80%)	10335 (1%)	117418
1-1, 3, 4, 5-1, 6, 8-1	All Above	1045122 (27%)	2767784 (71%)	90536 (2%)	3903542

Table 2
Districts With Genetic Outreach Clinics

DISTRICT	LOCATION	BLACK	WHITE	OTHER	TOTAL
1-2	Dalton	4924 (2%)	224483 (97%)	3198 (1%)	232605
2	Gainesville	19259 (6%)	278766 (92%)	6432 (2%)	3044624
5-2	Macon	155605 (37%)	257502 (92%)	4274 (1%)	417381
7	Columbus	136112 (41%)	188174 (57%)	7825 (2%)	332111
8-2	Albany	128391 (40%)	189102 (59%)	2934 (1%)	320427
9-1	Savannah	86228 (35%)	152513 (63%)	381 (2%)	242622
9-2	Waycross	62950 (23%)	205056 (76%)	3416 (1%)	271422
9-3	Brunswick	50029 (28%)	120791 (78%)	4862 (3%)	175682
10	Athens	57945 (21%)	215877 (79%)	4140 (2%)	277962
1-2,	All Above	7011443 (27%)	1832264 (71%)	40967 (2%)	2574674

Through contracts with the State of Georgia, two major tertiary care centers (Emory University, Atlanta and The Medical College of Georgia, Augusta) provide genetic medical services on a bimonthly basis at the 9 outreach clinics. The services are conducted by two board certified geneticists who generally bring along various clinical and support staff. Each outreach clinic is staffed by a genetic outreach nurse who coordinates the activities and patient management for each clinic.

Data in the tables reveal the following information: The 9 organized public health service genetics outreach clinics serve 40% of the total population of Georgia -- further broken down, into 27% black, 70% white and 2% other races.

The category of "other" in Georgia is not broken down by specific ethnic groups. The percent of Georgians in areas FIGURE 1 surrounding the tertiary care centers (42% of the total population of the state) reflects a racial composition of 30% black, 67% white and 3% other races. Of the remaining 18% of the population not served by an organized public health service genetic outreach clinic or being within tertiary center districts, 19% are black, 80% are white and 1% are other races. On the surface it would appear that minority groups are not disparately affected by the system or distribution of genetic services, but the black/white racial distribution within and between counties is an important consideration in arriving at conclusions regarding impact of services. The overall black population of Georgia is about 27% of the total population; however, the distribution of the black population varies considerably, not only as to wide geographic areas in the state; but, between adjacent counties as well. In some locations it is not uncommon to find one county with a black population 30% or greater while a bordering county has a black population of 5% or less. Of the districts not having a genetic outreach clinic, only District 1-1 (Rome) has a black population less than 20%. By percentage figures, it would appear that the Georgia "other" population is insignificant, however, there are "pockets" of "other" racial communities within counties that provide the greatest economic opportunities for their general welfare.

The lack of outreach clinics in the Rome (District 1-1) and Valdosta (District 9-1) regions represent differing policy rationales. In the Rome district there is no genetic outreach clinic because the district director did not see genetics as a high-priority health care need for that district. In the Valdosta region, neither Emory nor Medical College of Georgia outreach service providers conduct clinics in the subdistrict citing distance and time as a key factor. It is our understanding that residents of an unserved district find it difficult, if not impossible, to present at the clinics of neighboring districts. We have learned that some of the residents in the Valdosta district have received service through the Florida outreach system. This may be but a microcosm of a much larger, yet unreported, national problem. On the one hand, this as an example of how systems in place to serve the state's population present issues of access and justice in service delivery; but on the other hand, it also provides an example of interstate cooperation of public sector genetic services.

During the period July 1, 1991 through June 30, 1992, 2693 patients (1426 new and 1267 repeat) were seen in the 9 genetics outreach clinics (personal communications, Mary Ann Henson, Genetics Program Manager). The CORN Minimum Data Set Report: 1992 (published September 30, 1994) reveals the total number of patients served by the state (CORN Report Table 8.1) for this same period to be 9,544. This latter figure includes the patients served by the outreach clinics as well as patients drawn from the remaining 3,903,500 residents (approximately one-half the population of Georgia) living in areas not served by a genetic outreach clinic (Table 1). Based upon these data, it appears that the genetic outreach clinics account for approximately 30% of the total genetic activities supported by state or federal funding. The genetic clinics patient load does not generally include newborn screening patients. At present, there is no uniform system of data and record keeping in the genetic outreach system. Each genetic outreach location reports clinical activities in a somewhat individualized format. A uniform reporting format is under development by the program coordinator. In one Georgia district (7) the genetic outreach nurse is also the lead nurse for that district's early intervention program (EIP). After nearly a year in this unique position, she has a distinct impression that the early intervention program has increased referrals to the genetics clinic but there are no hard data supporting that impression. We have suggested the possibility of reviewing the intra-family service plans (IFSP) mandated by EIP directives as a potential means of assessing whether the EIP is creating more activity for the outreach clinic.

Newborn Screening: Functionally, and in part implied by law, there appear to be three "systems" embedded within the Georgia public health genetics services system: The newborn screening program under legislative mandate (O.C.G.A. 31-12-5) addresses metabolic disorders (O.C.G.A. 31-12-6) and hemoglobinopathies (O.C.G.A. 31-12-7) separately, targets sickle cell screening to voluntarily identified ethnic origins, states a broad purpose related to prevention of mental retardation (of which some of the conditions listed have no bearing on mental retardation), but empowers the Georgia Department of Human Resources with considerable latitude in developing a comprehensive state-wide medical genetics program.

Mary Ann Henson, Georgia Genetics Program Manager has put together a very comprehensive, easily understood genetic services manual on newborn screening. We are in the process of examining the newborn screening program in greater depth. It appears that the newborn screening network is served by two genetics counselors (one at each of the two tertiary care sites) who receive and interpret laboratory data and are responsible for dissemination of information

to other health care professionals (e.g., genetic outreach nurse or other public health system nurse) to follow through on testing and treatment initiation. To what extent counselor-to-patient encounters occur is, at this time, uncertain. We do know that the genetic outreach nurses with whom we have spoken do not regard or refer to themselves as genetic counselors, citing lack of sufficient training, blurred roles in the system and legal liability as factors. Clearly, states may be held liable for negligence in delivery of genetic medical services (*Marcel v. Louisiana State Department of Health and Human Resources*). In addition, some cases regarding medical malpractice in misdiagnosis or a hospital's failure to conduct screening or obtain an adequate blood sample never reach trial and are settled out of court. There are only unofficial records and word-of-mouth information regarding such cases.

A state-wide hemoglobinopathy program was initiated in Georgia in 1980. Georgia law provides for counseling regarding sickle cell anemia or trait to be furnished by county health departments at no cost to any person requesting counseling at no cost to the recipient (O.C.G.A. 31-12-7(c)); and, in fact, places an affirmative duty on the examining physician or the department (of health) to inform parents of children found to have sickle cell anemia or sickle cell trait that counseling regarding the nature of sickle cell anemia or sickle cell trait is available without cost (O.C.G.A. 31-12-7(b)). Legal research on this affirmative duty bears further investigation because it exposes the physician and/or the department to liability in negligence per se for omission to properly perform such duty. Our current information reveals that 68,615 initial screening tests for hemoglobinopathies were performed on an estimated 114,818 live births in Georgia and 182 cases were referred for confirmatory diagnosis and treatment. Of the 6,153 trait carriers identified, approximately 2/3 were counseled. Research efforts are underway to determine loss to follow-up and obtain more information on the nature of counseling and reasons why 1/3 of those identified did not receive counseling. The nature of state-sponsored activities as compared to private sector-sponsored activities in the sickle cell program is also under study in our current research. Apparently, even after many years of study, much misunderstanding remains in the realm of hemoglobinopathy disease management (Wright & Patton, 1990). Recognition of variations in genetic disorders is of prime importance to gaining a better understanding of diagnosis and management (Bowman & Murray, 1990).

FLORIDA

Florida's approach to the delivery of genetic services differs from that described for Georgia. The delivery of services in Florida involves a consultant-specialist cohesive team approach. The state's three medical schools supply genetics teams that provide specialty services under virtually identical, but geographically distinct, and legally separate contracts with the Florida Department of Health and Rehabilitative Services (HRS). Services include genetic testing and genetic counseling provided by teams consisting of medical geneticists and masters degree trained genetics counselors. The site of service provision is typically one of 22 clinics that exist within Florida Children's Medical Services (CMS) Program. CMS is a state program that represents a dramatic expansion of federal crippled children's programs. It provides care to children with chronic diseases and has financial eligibility criteria separate from (and more inclusive than) those established by Medicaid or other public programs. HRS develops contractual arrangements to provide continuing education for CMS nurses and this offers elective opportunities for genetics training for the on-site CMS staff.

Florida's genetic services are coordinated by the Department of Health and Rehabilitative services and delivered primarily through the auspices of the state's three allopathic medical colleges, which operate Regional Genetics/Endocrine Centers (Figure II), and the 20 Children's Medical Services (CMS) Offices throughout the state (Figure III). During calendar year 1991 this system served 3,642 prenatal patients. These patients included 272 from rural areas, 3,221 from urban areas, 16 from out of state and 133 for whom address was not reported. The primary reason for prenatal counseling was advanced maternal age (2,222) with other important reasons being risk of hemoglobinopathy (238), teratogen exposure (219), abnormal ultrasound (157), and family history of heritable disorder or defect (362). The most common procedure provided was amniocentesis (3008) with ultrasonography (421) and chronic villus sampling (231) as other common procedures. Most patients were seen by a non-M.D. Genetic Counselor (2,763) versus 225 treated by a M.D. Geneticist. No fetal abnormality was found in 3,221 with abnormalities found in 170 and 18 with uncertain findings and 3 with inadequate data. 296 patients declined testing after counseling and 109 were judged as "testing not indicated" by staff.

The system also provided services to 4,010 clinical genetics patients (1,886 new, 1,641 repeat and 483 status unknown). Clinical genetics patients included 2,596 whites, 670 blacks, 50 other known categories and 467 for whom no ethnic category was reported. 408 patients were known to be of Hispanic origin. 316 patients were aged 1-28 days,

460 were aged 29-364, 1,197 were aged 1-4 years, 614 were aged 5-9 years, 356 were aged 10-14 years, 199 were aged 15-19 years, 767 were aged 20-44 years, 46 were aged 45-64 and 55 were aged 65 and older. These patients included 1,887 males, 2,120 females and 3 of ambiguous gender. Rural residents accounted for 485 of these patients, 50 were from FIGURE I FIGURE III out of state and persons of unknown residence included 122 patients with the remaining 3,353 being urban Floridians.

Newborn Screening: Infant screening began in Florida with the passage of Section 383.14 Florida Statutes, requiring the State Board of Health to test all newborn for phenylketonuria (PKU). This legislation was amended in 1978 to include screening for other metabolic hereditary and congenital disorders including hypothyroidism, galactosemia, and maple syrup urine disease (MSUD) as well as PKU. This legislation also created an Infant Screening Advisory Council (ISAC) (consisting of consumer representatives, medical school representatives, pediatricians, and representatives of state agencies), and made provisions for follow-up, diagnosis and treatment of infants with abnormal screening results. The program was expanded again in 1984 to add screening for hearing impairment and birth defects and to establish a confidential computer registry (Consolidated Registry) to maintain information on these children. In 1985 MSUD screening was discontinued by the ISAC because 500,00 screenings had detected no true positive cases. In 1988 testing for hemoglobinopathies, including sickle cell disease was added to the program.

Currently, the Infant Screening Program is administratively located within the Florida Department of Health and Rehabilitative Services (HRS), Office of Children's Medical Services (CMS). The program has the following stated goals: (1) To assure that all infants born in Florida are screened and the testing is processed within two weeks of birth. (2) To assure that all affected infants receive appropriate confirmatory testing, counseling and treatment as soon as possible. (3) To provide physician consultation with other health care providers regarding the treatment and patient care recommendations. (4) To maintain a system of sound fiscal management of all public funds supporting the Screening Program. and (5) To provide a comprehensive educational program for the various health care providers.

The infant screening law states that HRS will maintain a confidential registry of cases, including information of importance for the provision of follow-up services to prevent mental retardation, to correct or ameliorate physical handicaps and for epidemiologic purposes. The law does not require informed consent, but instead requires that all infants be screened unless parents provide written objection to physicians or other authorities operating the screening program. All infants identified as in need of services are provided services regardless of income through an appropriate center. Regional Centers are located as follows: PKU and Galactosemia - Miami, Tampa and Gainesville; Hypothyroidism - Miami, St. Petersburg and Gainesville; Hematology Centers - Miami, Gainesville, Tampa, St. Petersburg, Orlando, Jacksonville, and Pensacola.

Local services are provided by CMS clinics and by county public health units (CHPUs). These include determination of eligibility for CMS services (an expanded version of the Federal Crippled Children's Program), coordination of care for children with metabolic, endocrine and hematologic diseases, and provision of prophylactic treatment for children with sickle cell disease.

Chapter IV -- Early Intervention Programs

In our initial contact with Mary Ann Henson, Georgia Genetics Program Manager, in November 1991 (11/15/91) she advised that we consider the potential impact of PL 99-457 -- the Education for the Handicapped Act Amendments of 1986 (20 USC 1400 et seq.), amending the Individuals with Disabilities Act Part H, a program designed to assist states in identifying developmental delays in pre-school (birth through age 2) children and to institute early intervention measures to best meet the needs of the child and assist in providing a smooth transition into their school years. The Department of Education promulgated its final rule (34 CFR 303) under authority of 20 USC 1471 (58 FR 40958, July 30, 1993) -- Early Intervention Program for Infants and Toddlers with Disabilities.

As perhaps obvious from this historical perspective, early intervention (EI) programs were in their formative to early operational phases during the time of our study. PL 99-457 expressed an enthusiastic agenda setting forth a 5 year deadline (1991) for states to meet requirements for continued federal funding to operate their EI programs. In order to meet continued funding requirements states had to have in place complex interagency operational systems meeting 14 minimal requirements described under 20 USCA 1476. Development of an individualized family service plan (IFSP)

plays a key role in meeting these requirements. By one account, only 14 states had applied for 4th year funding with the majority of states delaying submission of 4th year applications until spring of 1991, a year behind anticipated progress (Brown, 1991). Despite the neophytic status of EI programs throughout the time of our research, we found and describe here evidence forecasting the likelihood that progress in medical genetics will be used to meet EI goals.

GEORGIA

Georgia and Florida have both successfully met the requirements for a federal grant under PL 99-457 Part H. Georgia's program is called "Babies Can't Wait" and Early Intervention Family Support is the state's revenue source through which services for eligible children and their families are provided. Children who have an established risk of developmental delay (physical or mental) are eligible to receive services under this program. Details of the program have been published by the Georgia Department of Human Resources, Division of Public Health. IDEA, Part H: Babies Can't Wait/Early Intervention Program -- Program Standards, July 1, 1993 and Guidelines for Early Intervention Family Support Funds, July 1, 1993, provide comprehensive descriptions of operational goals, safeguards of confidentiality, and rights of families participating in the program. "IDEA" is an acronym for Individuals with Disabilities Education Act (20 USCA 1472). Early operations of this program in two health districts in Georgia were studied and described as part of our investigation (Woodson, 1993). Contradictory to some critics of our research, we highlight here the ways in which we find the result of this aspect of our investigation, supported by the opinions and findings of others, to support our contention that genetic diagnostic technologies will play an ever escalating role in Part H programs.

We analyzed the Babies Can't Wait descriptive literature and program policies and paid careful attention to the child tracking system identification form (Statewide Pilot 10/91) codifying high priority conditions used for making eligibility decisions. Of particular interest were the conditions relating to genetic causation listed on the form. In addition to the NBS program conditions, the tracking form listed Prader-Willi syndrome, Down's syndrome and muscular dystrophy. In addition, fragile X syndrome is cited as an established risk for developmental delay in the evaluation and assessment section of the Georgia DHR IDEA Part H Program Standards. The case analysis study conducted by Ms. Woodson, revealed that some children were referred to EI through the genetic outreach clinic located in that district. In another district, we learned that the Genetic Outreach Nurse was also the director of the EI program. In spite of these inter- and intra-agency connections, no data were available to establish purposeful interactions between the two programs at that time. We believe this will change with the advancement of both EI programs and the HGP. The Genetic Outreach program and EI program in Georgia are both under the Division of Public Health; this would appear to enhance the opportunity for collaborative effort.

Advanced technology made available through HGP research may result in redefining the scope of developmental delay thus impacting upon the scope of eligibility for EI services.

FLORIDA

In Florida, the Department of Health and Rehabilitative Services (HRS) developed its Part H entitlement program assigning Children's Medical Services as the principal agency of responsibility. In the Early phase of preparing for a Part H program, the Florida legislature codified its development and structure (FL ST 411.222). The Florida law on screening for metabolic disorders (FL ST 383.14), discussed earlier, expressly alludes to meeting requirements of PL 99-457. The Florida Part H program is described in a state HRS document, "Family Support Plan Protocol." It is obvious from these operational descriptions that EI programs in Florida, through inter-agency cooperation, take aggressive measures to identify infants at risk of developmental delay and actively to engage families in the construction of plans to use services to minimize the disadvantages faced by children with developmental delays as they progress toward their educational years.

SUPPORTING LITERATURE

Part H leaves up to the states how to define eligibility for services for children to be considered at risk (no established diagnosis and no demonstrative developmental delay). As genetic-based diagnostics is geared toward predicting risks, it seems logical that as technologies in this area become more sophisticated, more children at risk of developmental delay could be diagnosed as eligible for Part H services. The law automatically includes children with established diagnoses known to result in developmental delay (20 USCA 1472(1)(b)). Broad interpretation of legislative intent leads to myriad possibilities of conditions resulting in developmental delays (Shorkoff & Meisels, 1991). With this in

mind, we reviewed the literature to determine whether others were making connections between genetics and EI programs. In addition to the mental retardation causes of conditions screened for in many state NBS programs, there are other conditions that cause mental retardation and or physical developmental delay for which genetic technology-based diagnosis is a reality. Neurofibromatosis (causing disfiguration, speech disorders and cognitive defects) and fragile X syndrome (moderate to profound mental retardation in affected males and the possibility of borderline to mild mental retardation in females) are but two examples of such conditions mapped to specific gene loci (VanDyke & LinDyken, 1993). Possibilities such as these may form the foundation upon which to extend tests in NBS programs through molecular diagnostic techniques (Hoffman, 1994).

Some authorities implore counseling of individuals at risk for the neurofibromatosis-2 (NB2) gene with respect to availability of presymptomatic diagnostic genetic screening and offering of testing of children 2 to 8 years old in families considered to be at high risk for NB2 (Harsh et al., 1995). This recommendation falls within the EI infants and toddlers age described under Part H. Presently, there is no cure or effective treatment for this condition. Presymptomatic detection of disease for which there are no cures or effective treatments sparks heated debate within the genetics community. Early detection does not mean the child will have access to any treatment and it also raises questions generalizable to discrimination in health care coverage through insurance risk rating practices. The latter notion has become a thorny issue to the point where a spate of new state legislation attempts to severely limit the use of genetic information for health insurance policy determinations or other discriminatory purposes (McEwen & Reilly, 1992; Natowicz, Alper & Alpe, 1992; Rothenberg, 1995).

Although a hard-line stand on not testing/screening for conditions for which there is no cure or benefit to the individual is taken by some (Andrews, et al. -- "IOM Report"), 1994, an opposing argument may be made along lines of benefit to the parents in terms of making procreative decisions (Clayton, 1992). We hold that parents should be informed of childhood onset conditions that may be presymptomatically detected on the ground that it would be in the best interest of the child for the parents to make preparations for their special needs in advance rather than to be "surprised" as symptoms begin to develop. We believe the philosophy, if not the expressed intention of Part H supports our view on this matter. We understand and respect arguments contra to our position by those who point out that knowing adverse information in advance could be prejudicial to how the child may be treated by the family. We cannot argue that such would not be the case but we would ask why the child would not be treated "prejudicially" with heightened expressions of compassion and care rather than adverse expressions of rejection and disdain. We find, however, no evidence to support either view in any researched context. We do acknowledge that there are reported cases based on personal experience that provide information to be considered from parental points of view. For example, the case of a couple who had a PKU-affected but, because of careful care and monitoring, developmentally normal daughter and wished to have a prenatal test on a subsequent pregnancy to determine the chances of another PKU baby. There was no indication in the report that the parents did not love their daughter because of her potential affliction but were making decisions based upon emotional and economic factors that would result from having a second child with PKU (Elsas, 1990). Although there may be no currently available treatment or cure for conditions such as fragile X, neurofibromatosis or Duchenne muscular dystrophy, we forecast that EI programs will provide impetus for developing new ways to meet beneficial goals expressly stated in Part H for eligible children. If that should turn out to be the correct view, then controversy regarding early testing for these conditions would be moot and, perhaps as in the PKU case described above, other benefits may accrue.

Although NBS was discussed in greater detail in a separate section of this report, we include further discussion here as it is a system of EI. The conditions chosen to be screened in NBS programs, given variations from state to state, are generally justified on the basis of availability of effective treatment. Most NBS conditions would result in serious mental or physical disability if left untreated. We researched the literature to determine whether other conditions, even though treatment may not be presently available, are being considered or recommended to be included in either NBS or EI profiles. Around the time our research began, a report was published on a pilot program in NY State designed to screen post-pubertal mentally retarded males for fragile X syndrome (Nolin et al., 1992). The stated purpose of the program was to identify affected males and inform the females in their families who are at risk for inheriting the mutation and to counsel them that they may wish to determine their carrier status and consider that information in making reproductive decisions. In this process, 11 other chromosomal abnormalities were identified. The families were contacted by "local developmental disabilities personnel" and a genetic counselor. The authors of the report acknowledged that "lower functioning carrier females" may not have the capacity to even consider genetic counseling

let alone understand what it might reveal. A following article in the same issue of the journal reports on another fragile X screening program (Gabarron et al., 1992). That study, conducted in Spain, relies heavily upon cost-effectiveness for its justification, reporting that the cost (estimated \$12,740) of preventing a birth of one affected male was less than the average yearly cost of caring for a mentally retarded individual. The authors of the Spanish study predict that the diagnostic costs will be significantly less using direct DNA testing. Could it not be argued here that, under its stated purpose, the sovereign is essentially giving its citizens moral advice? Another comprehensive review on genetic testing for fragile X concludes that every child, male or female, with delayed cognitive abilities or who demonstrates symptoms of hyperactivity or autism should be offered DNA testing for fragile X (Rousseau, 1994).

The field of study linking genetic causation to cognitive/behavioral dysfunction is expanding. There is now evidence suggesting major locus dominant transmission in dyslexic families (Pennington, 1995). Pennington acknowledges that genetic consultation is not currently warranted in most cases of learning disabilities but predicts that once tightly linked markers are identified, they may be used to advantage for early intervention. Prenatal diagnosis of Klinefelter syndrome and other chromosomal abnormalities is already possible through fetal cell isolation from maternal circulation (Simpson & Elias, 1994). The National Institute of Child Health and Human Development encourages research on genetic causes of mental retardation (Gene Therapy for Disorders Causing Mental Retardation, NIH Guide to Grants and Contracts 22(3):20-22, 1993; Genetic Disorders Causing Mental Retardation, NIH Guide to Grants and Contracts 23(7):30-33.)

In the future we plan to continue research along these lines of inquiry and take our investigation further to identify and debate differences at state levels of genetic services operations to determine whether or how future planning for public sector genetic services are considering issues raised here. Casting no aspersions on the CORN and its many committee functions, it cannot be said that this organization and its composite national regions are considering critical issues in a carefully planned research context. Examination of the CORN reports produced thus far reveal that the organization is making serious efforts to categorize genetic services from an inventory perspective but falls far short of making any analysis as to policy decisions or trends. Without in depth investigation of programs, there is no way to detect the subtleties of routine operations. For example, we note that it is reported in the IOM report (Andrews et al., 1994) that Pennsylvania screens newborns for Duchenne muscular dystrophy. Upon checking with officials in the state's NBS program we learned that this is not done as a matter of routine but is, rather, a supplemental screening program conducted by an interested research specialist. This raises questions along the lines of what forces drive public sector genetics programs, who sets the services agenda, and would a supplementary (or pilot) program be subject to the same regulations as the mainstream program?

We believe, based upon our research findings and review of current literature, that products of rapidly advancing HGP research will not lie dormant while ethical debates proceed to forge the path ahead for just distribution and thoughtful incorporation of new technologies into medical practice. There has been a notable shift in marketing of biotechnological products wherein initial stages of research and development occur outside of commercial industry (academic and government research laboratories) and only at some commercially foreseeable (profit-forecasting) stage is the development taken over by the biotechnology industry (Silverman, 1995). Silverman predicts that the rate of commercial development of DNA diagnostics will not be impeded by on-going social issues debates. History supports the contention that a test available begs to be a test used. Cost of tests that, on the surface, appear to be expensive may be considered quite reasonable by couples who are willing to pay for an answer that they believe will be worth much more to their future emotional and financial standing (Silverman, 1995).

The information presented above supports our initial postulate that EI programs will look to genetic services at increasing rates to determine candidate eligibility even when that may be predicted prior to any overt symptomatology. In turn, the genetic services programs will use new DNA diagnostic technologies to fulfill such new demands. Although we have raised significant issues remaining to be resolved regarding how public sector genetic services will be impacted by EI programs and newly developed DNA diagnostics, specific answers can come only from carefully planned and thorough state-by-state case analyses.

Chapter V -- Ethical, Legal, and Social Issues

Our research objectives included analysis of ethical, legal and social issues, particularly those involving the concepts

of autonomy, justice, beneficence, and confidentiality of genetic information in light of rights, duties, and privileges currently established or suggested to be established by law and embraced in ethical codes of professional conduct. Commensurate with the beginning of the HGP and its continuing rapid progress, codes of ethics, both newly written and revised, by professional societies in clinical genetics reflect attempts to address various concerns raised by the rapidly expanding field of genetic research and the enigmas presented in control of genetic information. We examined the views expressed in the codes of ethics of three leading organizations: the National Society of Genetic Counselors (NSGC), the Council of Regional Networks (CORN) for Genetic Services, and the American Medical Association (AMA) Council on Ethical and Judicial Affairs, Current Opinions with Annotations (1994). The statements expressed in these codes of ethics are contrasted and compared to published studies on public attitudes and behavior related to genetic testing, genetic counseling and issues of information control. Our treatment of ethical and legal issues is by no means exhaustive but is intended to raise and debate questions yet unsettled by any measure either through research or applied policy analysis.

NCSG

A key question in our debate is to what extent views expressed by professionals in genetics align with views expressed by recipients or potential recipients of genetic medical services. The NSGC Code, January 1, 1992, is based on "relationships" (self, clients, colleagues, and society) and is written from an "ethic of care." The NSGC chose to use "client" rather than "patient" because the former implies empowerment while the latter conveys a more submissive attitude or role. Perhaps the primary component of the NSGC code is II item 3 -- "Enable their clients to make informed independent decisions, free of coercion, by providing or illuminating the necessary facts and clarifying the alternatives and anticipated consequences." Other than reiterating the interpretation that this section expresses a high value of respect for personal autonomy, self-determination and human dignity, little more is offered in the code's explication (Benkendorf et al., 1992). The explication is more of a restatement or elaboration of the code's rhetoric than a guide by example. The code is not intended to serve as the basis for disciplinary functions but is, rather, an expression of expectations of members of the profession. Although the term "nondirective" does not expressly appear in the NSGC code, the phrase "free of coercion" may be interpreted to imply non-directiveness in imparting information to clients. Certainly, the genetics profession(s) in general acknowledge(s) that to be completely nondirective is virtually impossible (Pencarhina et al., 1992).

Exactly what genetic counseling is and what purpose it serves have been issues under discussion for many years (Fost, 1992). It is doubtful that persons facing difficult decisions embrace the concept of neutrality in receiving information. It would be fair to say they are probably hoping for direction from experts. Nondirectiveness fosters a tension within the field of genetic counseling that is difficult for some of its members to resolve. That is, A MS degree genetic counselor may be in a position to act in a more "neutral" manner than a MD geneticist because their professional roles and images are quite different. Unlike the physician geneticist, the MS genetic counselor does not act in the capacity or have the image of care giver or healer. Consumer expectations from these two roles may therefore be quite different. This is not meant to imply that MS genetic counselors are not compassionate or caring. On the contrary, our conversations with these professionals gives us quite the opposite impression. Consumers as patients or clients expect expert advice from professionals -- advice that will not merely enable them but may even persuade them to make certain choices or justify choices they make. However, in this regard, the role professed to be taken by MS genetic counselors appears to be analogous to a view often criticized among technical/scientific experts -- persons who dispense technically accurate information but offer no moral guidance as to its use.

An interesting aspect of the MS genetic counseling profession is its governance. It is not officially regulated (by state or federal boards or agencies of legal authority) as to education and practice as is the case of many other health professionals dealing with human lives. This is not to say that the MS genetic counseling enterprise is poorly managed. On the contrary, in spite of this regulatory peculiarity, it appears to be well-governed through professional societies. This does, however, raise the question of to whom the MS genetic counselor is responsible and to what degree do they operate as independent professionals? Even more difficult to answer, is exactly what is genetic counseling and how is it evaluated in the scheme of health care services? Where will its role fit into the managed health care plans where outcome measures some objective form of "success." Should MS genetic counselors evaluate cases and provide information to geneticists MDs or other physicians rather than directly to clients? One expert confuses the issue by stating that counseling should be distinguished from advice but then proceeds to explain genetic counseling activities in terms of "advising" (Chadwick, 1993). Chadwick's expertise is in the areas of ethics, philosophy and law. The role

of genetic counselors from a medical sociologist's perspective is described as mopping up messes or doing the uncomfortable dirty work that others prefer to shun (Bosk, 1992). We ask whether MS genetic counselors put themselves in an impossible situation by espousing, on the one hand, a client-centered, nondirective approach; and, on the other hand, their role in "[P]articipating in activities necessary to bring about socially responsible change[s]" (item 2 IV, NSGC Code of Ethics) as well as "[s]erve as a source of reliable information and expert opinion for policy makers and public officials{.]" (item 3 IV, NSGC Code of Ethics). Individual (client) interests may be in conflict with the interests and values recognized as legitimate societal interests. (Fost, 1992). In terms of its meaning in the practice of law, "expert opinion" means persuasive (substitute "directive") information upon which conclusions are reached. How do MS genetic counselors propose to resolve this seeming conflict in their role as dispensers of "neutral" (value-free) information? Policy cannot be said to equate to autonomy. It appears from this code that respect for autonomy is presumed to be limited when its results would collide with societal needs and values. Societal needs and values as defined in operative policy differ from state to state. These differences, in fact, present ethical dilemmas by imposing restrictions on personal autonomy. Specifically, some states provide Medicaid-covered prenatal diagnostic services but disallow payment for termination of pregnancy for fetal anomaly (Weiner & Bernhard, 1990). How do genetic counselors resolve such a dilemma where autonomy does not, as a matter of practicality, include all choices for a subset of the population?

Near the beginning of our research, a study comparing perspectives of MS genetic counselors' to medical (MD) geneticists' was reported (Pencarinha et al., 1992). Among the differences between the two groups was the greater likelihood that MS counselors would not divulge information to family members at risk or to the spouse of a patient diagnosed with Huntington's disease where that patient does not want such information to be revealed. The tensions and ethical dilemmas in this case are palpable. Should respect for autonomy (the basic foundation principle of the NSGC Code of Ethics) supersede the broader principles of justice and possibly beneficence to third parties? That is to say, not telling others what could happen to them because of this illness is not only potentially damaging to the index patient (legal documents required for handling the affairs of an incompetent person must be executed while the person is legally competent) but may trespass upon the autonomy of potentially affected children of the patient by disallowing them to make informed choices (e.g., in the nature of procreation and future health care directives) about their own lives. There was no indication in the Pencarinha report whether the patients' choice was silently accepted by the counselor or whether discussion would ensue to allow such a decision to rest with full knowledge as to its potential consequences. It is questionable also as to whether a patient in this position has the legal capacity to make a decision about not telling relatives or whether the profound knowledge of the condition in fact impairs objective decision making. This latter issue is currently under legal debate with respect to capacity to execute wills, generally considered to be a low-level of competence, in persons affected with diseases that impair mental function, e.g., Alzheimer's disease, (Friedland & McMonagle, 1996). The Pencarinha report offers reasons why MS genetic counselors place such a high value on personal autonomy, based on differences in training in counseling and gender perspectives (most MS genetic counselors are female and most MD geneticists are male). The report concludes by acknowledging the views of others suggesting re-evaluation of value systems in medical genetics.

CORN

The CORN Code of Ethical Principles for Genetics Professionals -- April 1994 is based upon "responsibilities" with regard to patients and their families, society and members of the profession. The code expressly refers to "nondirective" counseling and providing information in a way that allows patients to "[m]ake independent decisions and give informed consent." While respect for autonomy and confidentiality are stated in the code, patients are encouraged to share pertinent information with relatives at risk. There is a brief glossary of some terms at the beginning of the document. The CORN code specifically states its responsibility to society in the expression: "[A]chieve appropriate balance between the rights of individuals and the need of public health in the use of genetic information." Here we see the notion that personal autonomy may not supersede societal needs and values. Both the NSGC and CORN codes have nearly identical statements in regard to protections against discrimination, except that the CORN code is silent as to the category of "age" in its list of generally recognizable suspect classes of persons identified in law as meeting criteria for legal protections against discriminatory practices. The CORN code goes further along these lines to include a statement on the responsibility of the profession to ensure that human rights are not trespassed on the basis of genetic characteristics. This latter point (actually the last statement in the code) raises interesting questions. Does this imply that the profession believes there is future possibility of genetically characterizing individuals on a sound scientific basis and that such could lead to historically repulsive abuses?

AMA

The AMA code is divided into sections of subject matter categories. Section 2.12 Genetic Counseling describes the ethical obligations of physicians engaged in genetic counseling, founded on the professional responsibility to "[p]rovide prospective parents with the basis for an informed decision for child bearing." Although the word "nondirective" does not appear in the rhetoric of this section, it is perhaps implied in precautions that physicians should avoid the imposition of personal values and moral judgments and the obligation to reveal personal conflicts and provide notice when a problem is detected so that patients may choose to seek further counseling if they so wish. The code does not express an affirmative obligation to suggest referral to a qualified professional. This section of the AMA code concludes with a discussion of "genetic selection" defined as "[T]he abortion or discard of a fetus or pre-embryo with a genetic abnormality[.]" and closes with a statement that selection on the basis of non-disease related characteristics would not be ethical. The term "pre-embryo" is itself in controversy and in question as to its meaning and legitimate use in either law or medicine. Although the AMA code states that it is generally permissible to engage in genetic selection to prevent genetic disease, it is not generally permissible to use genetic tests to exclude workers with genetic risks of disease from the workplace (2.132(1)) or for the purpose of predicting a person's predisposition for disease by the health insurance industry (2.135). The latter section goes on to suggest that separate records might be considered as a measure of protection to prevent inadvertent sending of medical information requested by insurance companies. However, the code suggests that the physician reveal the fact that such information has not been included in the information sent. There appears to be a conflict here. On the one hand, the intentional separation of genetic information from the remainder of the medical record is seen as a measure to protect confidentiality and privacy and avoid potential discriminatory uses of the information. On the other hand, revealing that such information exists and is being withheld implies that it may have some probative value to the insurance company and begins to dismantle the shield of intended protection to the patient.

COMPARATIVE ANALYSIS

The NSGC and CORN codes are similar more so to each other than either is to the AMA code. The AMA code was the only one of the three examined here that cited references as background and foundation for some of its text. These references are used to provide the basis of up-dating the code by incorporating contemporary views and suggested policies in the sections of the code. A question is raised as to whether there is or should be a common standard of ethics among the various genetics professions. The CORN code offers the widest possibility of commonality in ethical principles. The concern for arriving at an understanding of ethical values in medical genetics does not stop with the expressions contained in these three U.S.-based organizations. The World Medical Association (WMA) is busy fashioning its position on the issue of human genome data and genetic screening guidelines anticipated to be published in 1996 (Dickson, 1995). It will be interesting to see to what extent the three codes cited here have played in the WMA position.

SOCIAL AND LEGAL ISSUES

What are the standards for genetic counseling and how is its effectiveness measured? The purpose and scope of genetic counseling was defined by an ad hoc committee of the American Society of Human Genetics and reported in 1975 (Ad Hoc Com., 1975). This report focuses on the communications process of genetic counseling and sets out 5 norms or goals that enable a patient and/or a family to make autonomous decisions. The report identifies the role of the medical social workers and public health (recall the description and discussion of the Georgia genetics program, supra) nurses as a source of communications between the counseling group and the patient or family. Responding to legislative mandate: 42 USC 300v-1(a)1(c)(198881), the President's Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research issued A Report on the Ethical, Social and Legal Implications of Genetic Screening, Counseling and Education Programs (February 1983). This document continues to represent one of the most detailed, cogent and comprehensive discussions of the moral aspects of genetic counseling. The issue of non-directiveness is vigorously discussed in that report and debates raised then remain largely unresolved today. The process of genetic counseling is thought to be most effective when conducted by a multidisciplinary team, including a variety of genetic specialists, social workers, and other specialists who can best determine specific information to be conveyed to a family through an appropriately trained counselor (Kelly, 1986). Kelly raises the concern of nondirective counseling in cases where the object of the counseling lacks normal mental capacity or is emotionally unstable. This dilemma is echoed in a later report indicating the acceptability of directive genetic counseling in special circumstances (Fletcher & Wertz, 1990). The determination to suggest directive genetic counseling involves

assessment of risk (e.g., how great is the harm to others who may need to know) and advanced notice to the patient or relatives that directive counseling may be appropriate. No examples were offered to explicate this possibility. The process that would lead to the conclusion that directive counseling is appropriate is not explicated and raises a number of questions. Who decides what the "harm" is or would be that is significant enough to open the door to the suggestion of directive counseling. How much incompetence, decided by whom, on what standards, represents the threshold of opportunity to raise the possibility of directive counseling? Are records kept on the counseling process? Would sterilization of either the patient or relatives ever be included as a suggestion in the directive options? What do the options include where some of the relatives of the index patient also have less than normal mental capacity?

Raising these questions is not intended to imply that directive counseling is driven by some malevolent purpose but to note that the issue of avoiding propagation of the mentally incapacitated has been a recurring theme in the world's history and that this theme has not been completely eliminated from discussions of health policy. Indeed, Healthy People 2000, an often cited guideline for steps toward a healthier nation, supports providing preconception information to couples at risk for conceiving offspring with genetic disorders (Healthy People 2000 14.12, 1992).

To what extent will cultural values and perceptions be taken into consideration when issues of competency and comprehension arise? A well-written report on cultural issues in genetic counseling reveals that mentally retarded children may be desirable as compared to "normal" children in certain cultural settings (Marfatia, Puales-Morejon & Rapp, 1990). That report offers enlightenment on family dynamics in poverty where genetic concerns are among the least urgent of problems faced in daily life. In case studies detailing a variety of difficult situations, it is clear that the concepts of "illness" and "abnormality" are perceived differently among cultures. Some cultures are more concerned with morphological stigmata and view learning disabilities as less important. How does the counselor balance these values in a health care system where services are planned and designed for "populations" and not for ethnically and culturally diverse groups?

What do studies attempting to assess the impact of genetic counseling reveal? What do counselors expect from the process of their efforts? A study on sickle cell risks conducted in Virginia from 1970 to 1982 attempted to find clues to these questions as well as to other pertinent issues raised above (Neal-Cooper & Scott, 1988). In that study, couples at risk were counseled on probabilities of having children with either sickle cell anemia or trait. Details of the counseling were not provided. Outcome data were determinable on 25 couples. Fourteen of these couples produced 13 offspring with serious hemoglobinopathies prior to learning of their risk and 10 affected offspring (7 born to 11 couples who had no previous children) following risk counseling. The counseling was said to be carefully designed to be nondirective although couples were encouraged to participate. This suggests that nondirective counseling seemed to have little apparent impact in terms of changes in reproductive behavior. Notable in the data was the fact that the 13 affected offspring were produced by 9 couples who had the lowest pregnancy rate (44%) following counseling compared to the 7 couples with previous pregnancies (80%) and the 11 couples with no previous pregnancies (90%). Whether the outcome in the group with the 9 couples (13 affected children prior to counseling) reflects the impact of counseling or the impact of having had an affected child was not determined by the research.

The major goal of genetic counseling is said to be empowerment, enabling the client(s) to make decisions in light of information that potentially bears upon the decision-making process. If that is the case, can the "outcome" of this type of counseling be evaluated at all? Perhaps, outcome should be determined only in terms of informed autonomy. This is to say that risks may be assumed in spite of information which could have been interpreted as a daunting precaution to avoid having children. Whether this was the case in the Virginia study was not assessed. No determination was made as to how many of the pregnancies were planned either before or after the counseling process. As discussed by Marfatia (supra) and colleagues, many factors go into the final decision, including the intense desire of young couples to have a family despite risks of illness revealed to them through counseling or through experience. As revealed in the discussion of the Virginia study, the couples were fairly evenly split as to whether risk information would affect any future reproductive plans.

Where abortion becomes a reproductive option in terminating a pregnancy because of a detected genetic defect in the fetus, it is unclear whether that decision is based on concern for the quality of life of the potential child or made out of consideration of other circumstances involved in raising a child born with a genetic disorder. Some data have been analyzed along these lines in a study involving parents with children affected with cystic fibrosis(CF) where it was

determined that only 20% of parents of children would abort a CF-affected fetus (Wertz et al., 1991).

As Fost argues, the key question now is what genetic counseling should be not what it has been historically, with much of that analysis predating the impact of HGP research by many years (Fost, 1992). Why should the concept of nondirectiveness involving genetic issues be any more important than in other hard-to-arrive-at medical decisions? Suppose a patient pregnant with a fetus that will be severely affected by a genetic disorder asks advice from the geneticist or genetics counselor. Suppose also that this patient has a poor level of understanding of complex issues and has essentially no family support system. Is it fair, equitable, just, beneficent to provide this patient with neutral (value-free) information without also supplying a frank opinion of what she is facing? How can the client assess the implications of information saying that the onset of the disorder will be: 1) at birth, 2) at age 3-4, or 3) at age 25 or older? Can the genetic counselor in a capitated system provide the same respect for the patient's autonomy and at the same time address the conflicting interests of society in terms of minimizing the costs of health care? It may be argued that nondirective counseling is "value-free" information, is essentially the truth, or is "not deceitful," but, it is important to ask whether it contains hidden assumptions or hides something that may be of material value to the receiver. We believe that these continue to be cutting-edge issues to be explored in depth as the HGP continues to provide us with a panoply of challenges.