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PROGRESS REPORT

Impact of Human Genome Initiative-Derived Technology on Genetic Testing, Screening and Counseling: Cultural, Ethical and Legal Issues. DOE/NIH Grant #DE-FG02-92ER61396 Ralph W. Trottier, Ph.D., J.D. (PI)*, Lee A. Crandall, Ph.D. (Co-PI)**, Faye Cobb Hodgin, Ph.D., J.D.*, Mwalimu Imara, D. Min.*, Ray E. Moseley, Ph.D.**, David Phoenix, Dr. P.H.*, Delores Armotrading (Graduate Student)**, and Sherrill Lybrook (Graduate Student)*, Morehouse School of Medicine*, Atlanta, GA and University of Florida College of Medicine**, Gainesville, FL.

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ABSTRACT

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Genetic medical services provided by the Georgia Division of Public Health in two northern and two central districts are compared to services provided in a district in which a tertiary care facility is located. Genetics outreach public health nurses play key roles in Georgia's system of Children's Health Services Genetics Program, including significant roles as counselors and information sources on special needs social services and support organizations. Unique features of individual health districts,(e.g., the changing face of some rural communities in ethnocultural diversity and socioeconomic character), present new challenges to current and future genetics services delivery. Preparedness as to educational needs of both health professionals and the lay population is of foremost concern in light of the ever expanding knowledge and technology in medical genetics. Perspectives on genetics and an overview of services offered by a local private sector counselor are included for comparison to state supported services. The nature of the interactions which transpire between private and public genetic services resources in Georgia will be described. A special focus of this research includes issues associated with sickle cell disease newborn screening service delivery process in Georgia, with particular attention paid to patient follow-up and transition to primary care. Of particular interest to this focus is the problem of loss to follow-up in the current system. Critical factors in education and counseling of sickle cell patients and the expectations of expanding roles of primary care physicians are discussed. The Florida approach to the delivery of genetic services contrasts to the Georgia model by placing more emphasis on a consultant-specialist team approach. The state's three medical schools house genetics teams that provide specialty satellite services, under contract with the Department of Health and Rehabilitative Services, to 22 sites. Although some aspects of this system are similar to the tertiary care center genetic physician contracts for services in the Georgia health districts, there are no on-site genetics services personnel at the site of clinical services provided in Florida. The Florida services include genetic testing and counseling provided by teams that include medical geneticists and masters degree trained genetic counselors. Florida Children's Medical Services is a program representing a dramatic expansion of federal crippled children's programs. Unique aspects of this structure are discussed and compared to programs in other states within the southeast region. Ethical issues, related to the principle of justice, are discussed in terms of rural-urban differences in access to genetic services and the interrelationship of these differences to concepts of race, ethnicity, variable incidence of genetic diseases and level of genetic predisposition to multifactorial diseases. Legal concerns involve expanding liabilities in the realm of general medical practice, risk communication and issues surrounding the concept of informed consent in genetic medicine and genetic counseling.

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RATIONALE:

Rapidly advancing technology may threaten our ability to provide genetic services within existing public sector health systems. Patient confidentiality and justice with regard to access to services for minority and medically underserved populations must be maintained in the context of rapid technological changes. A comparison of two neighboring states that use considerably different systems to deliver genetic services can provide information on existing problems and may suggest areas of need for future policy studies.

PROJECT OBJECTIVES:

Description and comparison of laws, regulations, and delivery system characteristics affecting public sector genetic services in Florida and Georgia

Description and analysis of genetic medical services delivered by Public Health Departments of Florida and Georgia

Comparison of roles of various types of providers within the public sector systems of the two states

Description and assessment of policies and/or delivery system characteristics that adversely affect identifiable population subgroupings (e.g., rural populations, ethnic minorities)

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METHODS:

Information is being collected via review and analysis of relevant laws, regulations and contracts for services and detailed, structured, on-site interviews. To date, interviews have been conducted with: genetics outreach nurses and other staff at four Georgia health district genetics outreach sites, providers of urban sickle cell services, a private sector genetics counselor, public health genetics program administrators in both states, Florida's statewide sickle cell program administrator, a district health director who played a key role in the early development of Georgia's sickle cell newborn screening program, two university based masters trained genetic counselors who provide services in Florida through a contractual arrangement with the state, and a tertiary care center medical geneticist who is a state contractual service provider.

PRELIMINARY FINDINGS:

This summary presents brief discussions of five key problem areas using categories of indicators similar to those described in the "Behavioral Model of Health Services Utilization" (Andersen, 1968) as modified and expanded (Aday, Andersen & Flemming, 1980). Implications for further research conclude each area described. In addition, descriptions of Georgia and Florida public sector genetic services are provided including utilization data, and demographic and geographic perspectives.

Area 1: Access to and Entry into Services

Those barriers appearing most significantly at this preliminary phase of the study are as follows:

Unequal distribution of public health medical genetic services/clinics/personnel in districts of Georgia.

Reliance on referral specialist approach in Florida so that local services are available only on scheduled clinic dates.

Consumer geographic isolation and transportation concerns for those referred to one of the tertiary care centers for follow-up.

Lack of awareness and misunderstandings of medical genetic services.

Lack of comprehensive service coordination - need for social services assistance.

Insufficient information and referral services.

Ethnocultural, educational, linguistic, and socioeconomic constraints.

Sickle cell services are not generally a part of the genetics clinics -- instead are handled by specialty centers.

Bureaucratic policies and priorities

These preliminary findings suggest that the overriding factors that adversely affect access to and entry into the medical genetics delivery system are budget deficits, institutional priorities, and a general lack of a collaborative consortium of various factions involved to form a readily-accessible medical genetics delivery system.

IMPLICATIONS FOR FURTHER STUDY:

There is a need to define ideal model(s) of genetics services delivery system(s) that optimize equality of access to and entry into services.

Area 2: Continuity of Service and Treatment

Preliminary data indicate that service coordination and continuity of care are sometimes fragmented due in part to the various independent inter and intra-institutional factions that influence and/or set policies for medical genetic services. Preliminary findings suggest common barriers to continuity of service are as follows:

Inadequate procedures for tracking and follow-up of consumers -- may reflect limited staff available to follow through.

Gaps in care of consumers during transition from childhood to adulthood.

Medical health needs and medical genetic needs perceived as independent and allocated to different public health units -- reflects a need for integration of genetics into primary and preventive medical care.

Gaps in information dissemination and referral networks among various providers in the system.

These preliminary findings suggest that the overriding factor creating a barrier to continuity of care is a lack of comprehensive coordination of medical genetic services.

IMPLICATIONS FOR FURTHER STUDY:

There is a need for development of guidelines to establish a comprehensive program that documents and tracks the consumer after the initial visit throughout the delivery system, promotes continuity of care, provides for information dissemination among providers, and protects consumer privacy/confidentiality/autonomy.

Area 3: Scope of Services Available

Technological potential to provide more comprehensive services for children, adolescents, adults with genetic disabilities, and their families requires a new paradigm of genetics. Preliminary data appear to indicate that an awareness of the need for an integrated services approach with well-trained resource persons is increasing among public health providers but identify common barriers to offering a broader scope of services as:

Limited medical genetic services for the adult.

Lack of social services and other support resources.

Limited number of scheduled genetic clinics -- organized genetics outreach clinics see patients only on specified dates (one day every other month in Georgia). Medical geneticists provide service by contractual agreements with the states.

Fragmented services for preconception, prenatal, natal, postnatal consumers -- aside from the newborn screening program, it is not clear to what extent and by whom patients are counseled on matters pertaining to genetics.

Primary factors emerging from preliminary data and significantly impacting the scope of services available in the public health sector include: budget constraints, priorities established by health authorities at state, district, and local levels, and assignment of genetics outreach nurses to duties other than their roles in genetic services.

IMPLICATIONS FOR FURTHER STUDY:

There is a need to design genetic services delivery models to consider coordination of genetics with other medical services, provide mechanisms for service evaluation, and have the flexibility to adapt to advances in genetic medicine.

AREA 4: Information Dissemination

Preliminary findings indicate that a comprehensive system of information collection, development, and dissemination concerning medical genetics and delivery services to both medical and lay communities is needed to meet the challenges expected to present with the Human Genome Project. Data suggest that insufficient knowledge, lack of information or misinformation are major barriers fostering reluctance by the provider to ensure comprehensive genetic services and a reluctance or non-awareness by the consumer to use these services. Our data to date suggest other barriers to knowledge and information dissemination are as follows:

Lack of a systematic and standardized method of data collection, analysis, and dissemination to evaluate medical genetic services.

Insufficient numbers of medical geneticists, genetic counselors, and genetic outreach nurses to meet consumer service needs.

Insufficient number of professional continuing educational programs for genetic outreach nurses addressing their specific learning and training needs in medical genetics.

Lack of community outreach and educational programs.

Inadequate referral linkages in medical genetics network of consumer, provider, institutions.

Communication and information gaps between and among the various components of the medical genetics delivery system.

IMPLICATIONS FOR FURTHER STUDY:

There is a need to develop information network systems with feedback - control - evaluation loops that would provide for information dissemination and educational updates to the public and health professionals.

AREA 5: Cultural Appropriateness, Sensitivity and Competency

There are numerous challenges posed by the many cultures and religions represented in Georgia and Florida coupled with increasing immigration particularly of Hispanic and Asian populations. Findings suggest common barriers to basic health care services and genetic services are as follows:

Cultural and language difficulties between consumer and genetic service provider create a gap in understanding vital information.

Lack of an organized system to provide interpreters and translators for those with limited English-speaking skills.

Limited learner-specific consumer teaching materials in genetics -- reflects need for understandability at various reading levels and skillful use of visual aids to enhance understanding.

Differences in cultural expectations, health beliefs, and value systems which are sometimes incongruent between consumer and provider -- reflects need for greater understanding, tolerance and respect for cultural diversity.

Dearth of ethnocultural diversity of staff in genetic services delivery systems.

Two primary factors appear to emerge as significantly impacting the delivery of genetic services to a culturally diverse population: first, budget constraints imposed on genetic services due to lack of funding or institutional policy priorities, and secondly, limited continuing educational programs for genetic services providers that meet their expressed need for greater awareness of cultural diversity issues and effective clinical interactional approaches.

IMPLICATIONS FOR FURTHER STUDY:

There is a need to propose policies that address comprehensive strategy to include the elements of funding, priorities in services, and needs assessment of consumer-genetic provider (e.g., Genetic outreach nurse) and take into consideration the increasing multicultural issues in genetic services.

Georgia Public Health Genetics System

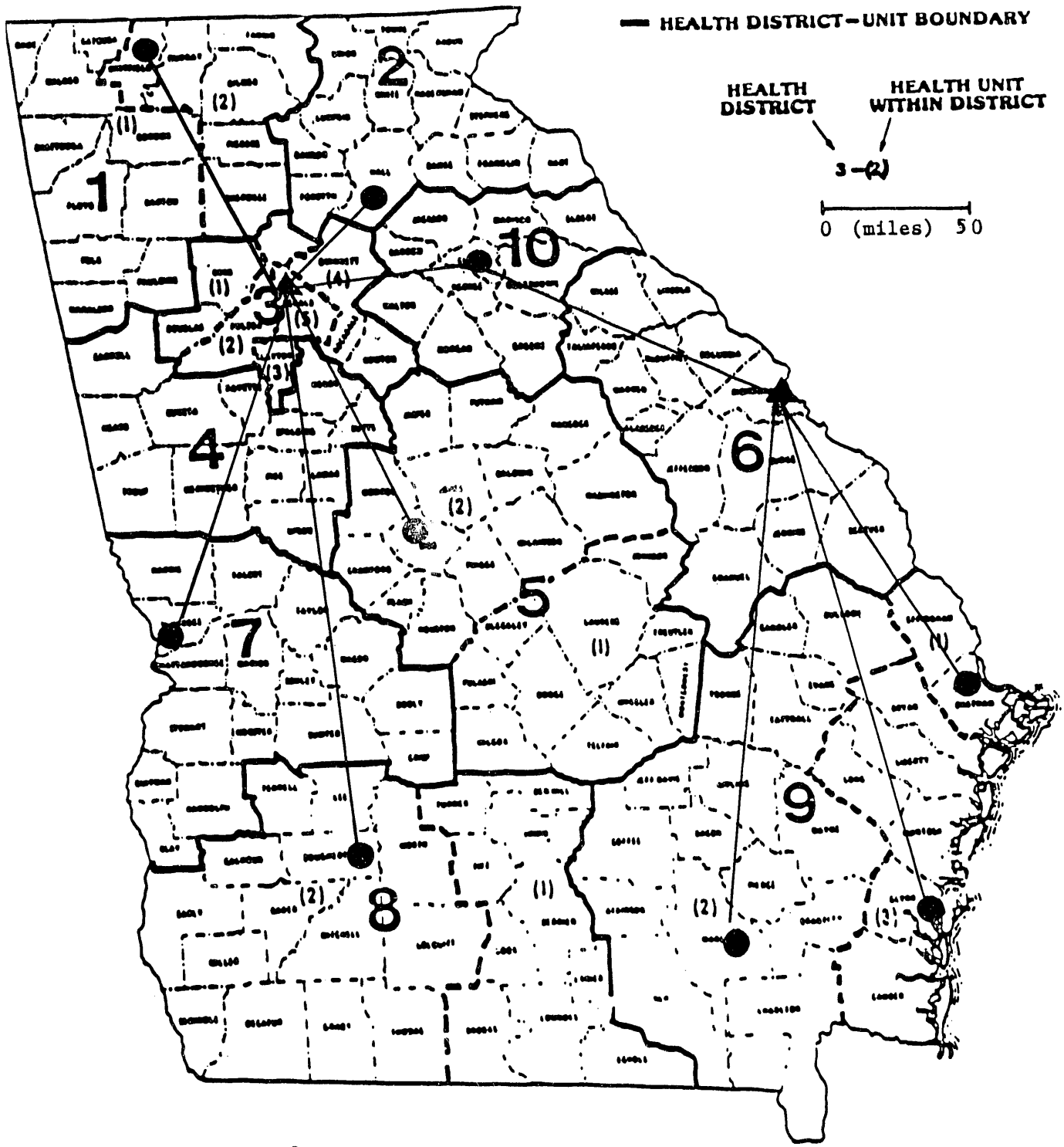
Figure I shows health district divisions of the 159 counties in Georgia. Each district and district subunit has a health department office. Through contracts with the state, two major tertiary care centers (Emory University and The Medical College of Georgia) provide genetic medical services on a bimonthly basis at 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 permanently staffed by a genetic outreach nurse who coordinates the activities and patient management for each clinic.

The 9 organized public health service genetics outreach clinics serve 40% of the total population of Georgia -- 27% black, 70% white and 2% other races. The percent of Georgians in areas 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. 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, but there are "pockets"of "other" racial communities within several counties that may require specialized genetic services.

During the period July 1, 1991 through June 30, 1992, 2,693 patients (1,426 new and 1,267 repeat) were seen in the 9 genetics outreach clinics. These figures do not include any information regarding the approximately 3,903,500 residents living in areas not served by a genetic outreach clinic. This is not meant to imply that these residents are without service but, rather, to convey information relative to a gap in record-keeping systems. The genetic clinics patient load does not include newborn screening patients.

Functionally, and in part implied by law, there appear to be three "systems within Georgia public health genetics services. The newborn screening program addresses metabolic disorders and hemoglobinopathies separately, targets sickle cell screening to voluntarily identified ethnic origins, states a broad purpose related

FIGURE I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Legend:
Tertiary care genetic service providers
Public Health Service Genetic Outreach Clinics

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. Apparently 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. 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.

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 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. 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 Public Health Genetics System

The Florida research team has taken a slightly different approach to data collection because the Florida 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 contract 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. Services are delivered with a periodicity that ranges from quarterly day-long clinics to monthly day-long clinics depending on the resident population size and demand for services at sites. The site of service provision is typically one of 22 clinics that exist within Florida Children's Medical Services (CMS). 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 those established by Medicaid or other public programs. HRS contracts for continuing education for CMS nurses and this offers elective opportunities for genetics training for the on-site CMS staff.

The Florida infant screening program began in 1965 with PKU screening and currently screens all newborns for hypothyroidism, galactosemia and hemoglobinopathies (particularly sickle cell disease). Maple syrup urine disease was part of the screen from 1978-85 but was discontinued after 500,000 births produced no true positive tests.

The Florida genetics services system provided clinical genetics services to 4010 patients in 1991, (1,886 new and 1,664 repeat). Of the 3,793 patients whose race was recorded, 80% were White, 18% Black, 1% Asian/Pacific islander, and 1% were of other ethnic groups. Ten percent of all patients were of Hispanic origin. Twelve percent of patients resided in rural areas. In addition, the program provided prenatal services to 3,642 persons (70% White, 14% Black, 2% Asian/Pacific Islander, $<1\%$ other). Sixteen percent of these patients were of Hispanic origin and 7% lived in rural areas of the state.

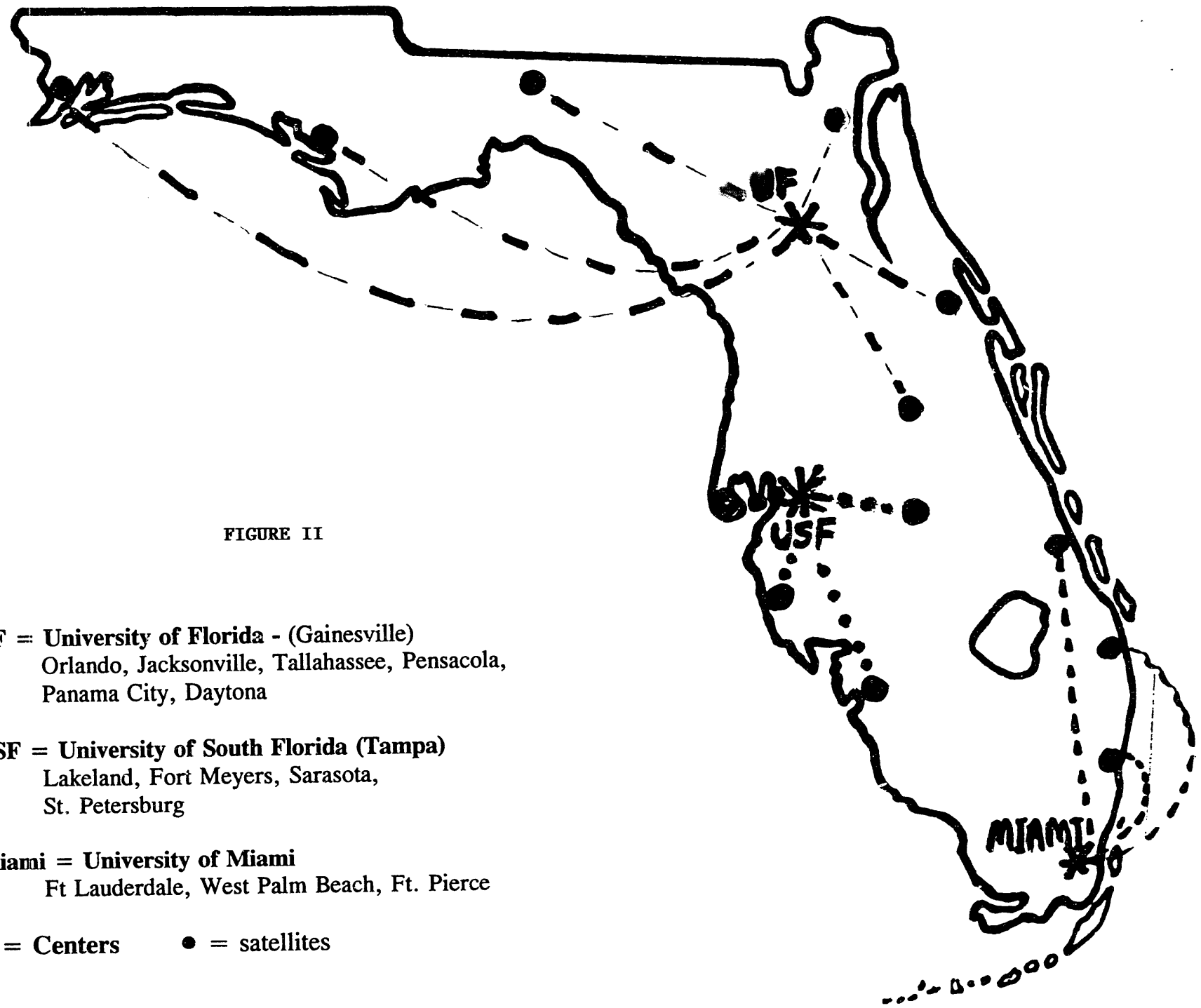

FIGURE II

UF = University of Florida - (Gainesville)
Orlando, Jacksonville, Tallahassee, Pensacola,
Panama City, Daytona

USF = University of South Florida (Tampa)
Lakeland, Fort Meyers, Sarasota,
St. Petersburg

Miami = University of Miami
Ft Lauderdale, West Palm Beach, Ft. Pierce

* = **Centers** ● = satellites

END

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