

**Guidelines for Clinical Genetic Services  
for the Public's Health**

**Council of Regional Networks  
for Genetic Services**



**GUIDELINES FOR CLINICAL GENETIC SERVICES**  
**FOR THE**  
**PUBLIC'S HEALTH**

**FIRST EDITION**

Council of Regional Networks for Genetic Services

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### TABLE OF CONTENTS

PURPOSE .....	1
BACKGROUND INFORMATION FOR GENETIC SERVICES AND THE PUBLIC HEALTH .....	3
I. THE CHANGING FACE OF HUMAN DISEASE .....	3
II. GENETICS AND THE HUMAN LIFE CYCLE .....	3
A. PRENATAL .....	3
B. PERINATAL .....	4
C. CHILDHOOD/ADOLESCENCE .....	5
D. ADULTHOOD .....	6
III. IMPLICATIONS OF HUMAN GENOME RESEARCH FOR DELIVERY OF GENETIC SERVICES .....	7
IV. PUBLIC AWARENESS OF GENETICS: THE CHALLENGE FOR PUBLIC HEALTH .....	7
 GUIDELINES FOR CLINICAL GENETIC SERVICES FOR THE PUBLIC'S HEALTH .....	 13
I. ORGANIZATION AND ADMINISTRATION .....	13
II. PREVENTION .....	16
III. SERVICES .....	18
IV. RESEARCH .....	28
V. EDUCATION .....	29
VI. DOCUMENTATION OF NEEDS AND SERVICES .....	31
VII. FUNDING .....	33
 AUTHORS .....	 35
 REFERENCES .....	 37
 APPENDICES .....	 39
A. CORN AND THE REGIONAL NETWORKS .....	41
B. CALCULATIONS .....	45
C. DESCRIPTIONS OF GENETIC PROFESSIONALS .....	47
D. DEFINITIONS OF SALIENT TERMS .....	53
E. LIST OF PUBLISHED STATEMENTS ON HEALTH SUPERVISION FOR CHILDREN WITH GENETIC DISORDERS, DEVELOPED BY AMERICAN ACADEMY OF PEDIATRICS, COMMITTEE ON GENETICS .....	55

# **GUIDELINES FOR CLINICAL GENETIC SERVICES FOR THE PUBLIC'S HEALTH**

## **PURPOSE**

The purpose of this document is to provide state and territorial public health agencies with an outline of suggested components for a genetic services system. Individual states and territories vary considerably in their genetic service needs. Therefore, this document has been designed to be used as a framework by local communities and agencies in developing their own comprehensive genetics plan.

Public health agencies are responsible for: 1) collecting and analyzing data to identify problems and community needs; 2) setting health goals, and identifying and mobilizing the resources to achieve these goals; and, 3) assuring quality and access to services to those in need of these services. This document serves as a resource tool for background on genetics and public health implications, data collection, and funding sources. The document emphasizes service delivery with descriptive backgrounds of types of genetic services, personnel, and quality assurance references.

Genetics is a rapidly developing discipline which is providing us with the knowledge to: 1) prevent the occurrence of many birth defects; 2) treat the sequelae of genetic disorders; and, 3) decrease the burden of chronically disabling diseases such as cancers, diabetes, and heart disease. While the lives and health of thousands of newborns have been saved by successful population based screening for inherited metabolic disorders, the many other ways in which genetic services preserve health and prevent suffering have yet to enter the domain of public health (1) and the awareness of primary care providers. Genetic services exist to help patients and families understand their specific genetic disease or risk thereof within the context of their own psychosocial, ethnocultural background.

**The ultimate goal of genetics is to reduce mortality and morbidity, and to alleviate suffering associated with genetic/congenital disorders.**

These guidelines provide a framework to develop a state genetic system and are not meant to provide guidance about the practice of medical genetics. The guidelines are presented in seven parts: 1) an organizational and administrative structure, 2) discussion on the preventive potential of genetics, 3) types of family, population, and laboratory services, 4) research, 5) education, 6) documentation of needs and services, and 7) funding sources for services. In addition, these guidelines address formal qualifications of genetics staff, the need for privacy and confidentiality, and other issues pertaining to the provision of genetic services.

Other professional genetics organizations, such as the American College of Medical Genetics, the American Board of Genetic Counseling, the American Academy of Pediatrics, and others are

developing specific practice guidelines which address management protocols for individual disorders or groups of disorders, such as Down syndrome, mental retardation, stillbirths, breast cancer testing and counseling, and the approach to the dysmorphic child. (2 and Appendix E) Examples of some of these are also presented in this publication and were reported upon at the conference on Developing Guidelines for the Public's Health in Washington DC on February 16 & 17, 1996 (Conference Proceedings available at no charge from the National Maternal and Child Health Clearinghouse, 703-821-8955 ext. 317).

Health care delivery is changing rapidly, particularly in the rise of for-profit service delivery and managed care organizations. Resources are scarce and genetics professionals have less time to devote to supportive communication to the patients and families they serve. Public health agencies will play a critical role in overseeing these changes and assuring access to quality genetics care for their populations. The Council of Regional Networks for Genetic Services (CORN) recommends that each state/territorial public health agency become acquainted with its own Regional Genetics Network as well as with CORN. These networks are resources for each state or territorial public health agency (Appendix A).

## **BACKGROUND INFORMATION FOR GENETIC SERVICES AND THE PUBLIC'S HEALTH**

### **I. THE CHANGING FACE OF HUMAN DISEASE**

Health care during the first two-thirds of the 20th century was characterized by a significant reduction in the occurrence of infectious diseases around the world. Well-organized, coordinated, and monitored immunization and pasteurization programs as well as improved nutrition have resulted in a 25-fold reduction in childhood mortality (3).

In developed countries, epidemics of contagious disease have been replaced as leading causes of mortality and morbidity and major consumers of health care resources by congenital malformations, developmental and learning disabilities, and common chronic disease of adulthood and aging. In the U.S., congenital malformations are now the first cause of death in infants under 12 months, the second after injuries in toddlers and young children. Our understanding of the genetic basis of congenital abnormalities and serious childhood disease is growing rapidly and promises to offer solutions to this new public health challenge.

At the same time, dramatic breakthroughs are taking place in the recognition of genes that contribute to common adult-onset disorders such as cancer, diabetes, and heart disease. The task now is to translate this new knowledge of genetics into actions that will improve the public's health.

It is clear that effective implementation of collaborative genetic and public health initiatives can spare the health and lives of tens of thousands of children and adults each year.

### **II. GENETICS AND THE HUMAN LIFE CYCLE**

The preventive potential of medical genetics extends to all branches of medicine at all stages of the life cycle (4).

#### **A. PRENATAL**

1. A carefully explored family, medical and obstetric history in couples contemplating pregnancy can identify many factors prior to conception, which if appropriately managed, can prevent an unfavorable outcome of pregnancy.
2. Intrauterine exposure to alcohol, smoking, cocaine, and other hazardous agents increases the risk for physical and developmental disabilities. Fetal Alcohol Syndrome affects an estimated 7000 infants each year in the U.S. (5). Smoking is associated with small for gestational age infants and cocaine causes placental abruption and prematurity.

Well-coordinated public educational efforts targeted to women of childbearing age have the potential for saving thousands of children and millions of dollars.

**About 7000 newborns are born each year in the U.S. with Fetal Alcohol Syndrome. Intrauterine exposure to alcohol is totally preventable.**

3. Maternal folic acid supplementation prior to and during the pregnancy may prevent the occurrence of open neural tube defects (spina bifida) in approximately 2000 newborns per year. This represents minimal savings of  $2000 \times \$10,000 = \$20,000,000$  per year (6 and Appendix B-1).
4. Maternal serum multiple marker screening for specific birth defects identifies an estimated 5% of pregnancies at higher risk (approximately 200,000 per year in the U.S.), facilitates their appropriate management, and prevents unnecessary complications of labor and delivery (7).
5. Screening for fetal chromosome abnormalities in the estimated 6% (or 240,000) pregnant women each year who are age  $\geq 35$  years enables parents not only to make informed decisions, but to plan for timely and effective intervention for affected infants (estimated 5000/year) (Appendix B-2).
6. Prenatal monitoring and management of maternal diseases associated with risk to the fetus result in improved pregnancy outcome. Well-documented examples include the prevention of: 1) mental retardation in offspring of mothers with PKU; 2) congenital malformations and/or metabolic compromise in offspring of diabetic mothers; and, 3) complications in offspring of mothers with prenatal infections (TORCH, HIV, etc.).
7. Selected use of fetal ultrasound identifies abnormalities, enables appropriate management, and optimizes pregnancy outcome.

**B. PERINATAL**

1. Approximately 2-4% of infants (80,000-160,000 per year) are born with birth defects

that have serious medical or surgical implications. Many of these birth defects are preventable. About one third of all children hospitalized in tertiary care medical centers have genetic disorders.

2. Newborn screening for inherited metabolic disorders identifies 3000 infants each year in the U.S. who are born with diseases such as PKU, hypothyroidism, galactosemia, sickle cell disease, and thalassemia. Rapid detection leads to appropriate treatment and prevents mental retardation, physical disability, and death. For example, special protein-restricted diets instituted soon after birth prevent irreversible mental retardation in infants with PKU. Similarly, recent studies have demonstrated that penicillin prophylaxis saves the lives of infants with sickle cell disease.

**Newborn screening spares the lives and/or health of about 3000 infants in the US each year. (8)**

3. Birth defect surveillance systems in the states and territories can obtain baseline information to monitor changes in the incidence or prevalence of specific types of birth defects in specific locations.

### **C. CHILDHOOD/ADOLESCENCE**

1. Approximately 3% of school-aged children are cognitively disadvantaged. An additional number have learning, attention, or behavioral and/or emotional difficulties. Early provision of organized, well-coordinated, family-focused services for children with special needs prevents later complications in these affected children. Case management is an important component of such services.

**Early recognition and provision of organized, well-coordinated services for children with special needs (~3% of the childhood population) prevents later complications in these affected children.**

2. Common genetic disorders such as mental illness, diabetes, and metabolic disorders appear in childhood and adolescence.
3. **Mass education about alcohol and other drug abuse, about contraception and pregnancy planning can prevent unfavorable consequences in thousands of future offspring.**

## D. ADULTHOOD

1. Genetic issues associated with the childbearing years include pregnancy losses, infertility and pregnancies at risk for unfavorable outcome. Appropriately designed population screening to detect carriers of serious genetic disorders such as Tay-Sachs disease (1 in 30 Ashkenazi Jews are carriers) enables couples to make informed reproductive decisions.
2. Adulthood is associated with the appearance of common disorders of great public health significance including hypertension, heart disease, diabetes, and cancers, all of which have a genetic component. For example, the recent identification of genes which, when mutated, increase the risk for breast/ovarian or colorectal cancers, offers the possibility in the future of presymptomatic screening for some of these diseases, which have a cumulative lifetime risk for almost 20% of the population (9, 10).
3. Mental illness including schizophrenia and manic-depression have genetic components and the search is underway to identify the genes involved.
  - **Cumulative lifetime risk for breast cancer in all women ~1 in 8 (10)**
  - **Mutation in BRCA1 gene in (non-Jewish) young women ~1 in 800**
  - **Mutation in BRCA1 gene in (Jewish) young women ~1 in 107**
4. Late onset and degenerative diseases with known genetic determinants include Huntington and Alzheimer diseases.

The dramatic breakthroughs in genetic technology and the resulting expansion of molecular diagnostics make it critically important that we pay attention to cultural differences, quality assurance at all levels, and the active involvement of consumers and families in all genetically related services and deliberations (11). There are important ethical issues to be addressed as screening for late onset and degenerative diseases becomes available to the public.

Thus, the medical, economic, and social impact of genetically determined disorders is already enormous and their psycho-emotional implications are currently unfathomable. The potential for preventive management, and alleviation of suffering is already a public health issue of significant magnitude. It pales, however, in comparison to the increased needs anticipated within the next few years.

### **III. IMPLICATIONS OF HUMAN GENOME RESEARCH FOR DELIVERY OF GENETIC SERVICES**

In the immediate future, the Human Genome Project (HGP) will result in the identification of an increasing number of disease-causing genes leading to better risk assessment and diagnosis for thousands of genetic disorders. Strategies for prevention, management, and treatment of genetic diseases will impact ever larger numbers of individuals, families, and populations.

No adequate infrastructure exists to process the flood of knowledge 'trickling down' from the HGP to the professional and lay public. Yet the anticipated need and demand for genetic services will undergo a significant expansion within the next three to five years as awareness of the new genetic information increases. Millions of dollars are being directed toward the support of human genome research and its ethical, legal, and social implications. It is clear that similar support is already essential for training and service delivery, and for quality improvement (QI) of genetic services for the public's health. This must include the availability of specific, appropriately interpreted information about relevant issues to all patients and families who need it.

**Clinical guidelines are needed to define clearly the quality of care delivered by genetic service providers, such as clinical and laboratory geneticists and genetic counselors. In addition, primary care providers such as those in obstetrics, pediatrics, internal medicine and family practice, as well as specialists in areas such as oncology, surgery, and neurology, may need guidance if they are to participate in the initial workup and ongoing management of patients and families affected by or at risk for genetic disease.**

### **IV. PUBLIC AWARENESS OF GENETICS: THE CHALLENGE FOR PUBLIC HEALTH**

The media quickly recognized that genetics-related issues have an impact on a broad segment of the population. At the same time, it became obvious that genetic issues are public health issues. Rarely a month passes without a dramatic discovery in medical genetics. Such breakthroughs raise hopes for cures in many affected individuals and their families. Hence the crucial need for timely and accurate information about the significance of each discovery for the treatment of human disease.

Unfortunately, the current level of understanding and appreciation of the importance of genetic disorders by many health care providers has lagged behind the recent explosion of knowledge due to the following factors:

1. Genetic disorders, while cumulatively common, are individually rare, and therefore, individual practitioners may encounter only a few affected families during a lifetime of experience.

2. Traditionally, modern medicine is oriented toward the individual while the specialty of genetics requires consideration of the extended family as a unit.

**The unit of investigation in clinical genetics is not only the individual patient but also the family.**

3. Genetic disorders are usually permanent and often accompanied by chronic medical and psychosocial problems. They are occasionally disfiguring and perceived as stigmatizing by families and society. Patients and families are frequently reluctant to discuss their concern with their own family members and with their health care providers.
4. Genetic evaluations (e.g. pedigree analysis, cytogenetics, DNA testing) of individuals and families are complex and highly technical procedures for which few physicians or other health care providers are adequately trained to perform or interpret.
5. Genetic evaluations are time-consuming. When they involve multiple family members who may be geographically dispersed, they are logistically complicated.
6. Population screening (e.g. prenatal, newborn, carrier screening) are usually developed and implemented by state agencies with little involvement of primary health care providers.
7. Genetic services which are still frequently documented under the label genetic 'counseling' are rarely seen as essential by health care providers or third party payers. Few practitioners are aware of the reasoning behind accurate and appropriate referrals for genetic services.
8. Documentation of the benefits of genetic services, apart from newborn screening activities, has been difficult and incomplete, hence their importance is not recognized.
9. Although genetics has meaningful potential for the prevention of birth defects and other disorders, prevention is equated by many with elective abortion of handicapped children alone.

Clearly, genetics issues have achieved public health dimensions. The genetics and public health communities must collaborate in educating professionals and the general public about the impact of genetic issues regarding the health of the general population. Such efforts entail defining the role of public health agencies in facilitating access to genetic services for all families and populations.

Each state and territory should have a plan for an organized and well-documented system of genetic services based on a prior needs assessment as described in sections IC and VI. The following should be addressed in developing these individual plans as described in detail in subsequent sections of this document.



- I. Organization and Administration**
- II. Prevention**
- III. Services**
- IV. Research**
- V. Education**
- VI. Data Collection and Documentation**
- VII. Funding**



## **GUIDELINES FOR CLINICAL GENETIC SERVICES FOR THE PUBLIC'S HEALTH**

### **I. ORGANIZATION AND ADMINISTRATION**

**A. State/Territorial Genetics Coordinator/Educator.** Each state should identify a genetics unit or, at least, a full-time genetics coordinator/educator with a background in service delivery, genetics, and public health issues. The responsibilities of the coordinator should include:

1. a) Facilitating communication within the genetics community providing existing genetic services in the state/territory.  
b) Maintaining linkages between them, consumers and all relevant components of the state/territory Department of Health.
2. Familiarity with all aspects of clinical and laboratory components of genetic services including:
  - a) prevention
  - b) dissemination of information (training and education programs)
  - c) needs and resources
  - d) mechanisms of reimbursement
3. Understanding how genetic services are distributed within their state and promoting the accessibility of these services to all who need them.
4. Identifying needs for additional genetic services in their state.
5. Understanding existing data collection programs and addressing additional needs.
6. Monitoring state legislation and regulatory efforts directed at genetic issues.
7. Familiarity with recognized professional standards for clinical and laboratory personnel, facilities, and genetic services.
8. Monitoring all contracts related to state-funded genetic services.
9. Collaborating closely with the State/Territorial Genetics Advisory Council (see section IB below).

**B. State/Territorial Genetics Advisory Council.** Each state/territory should develop mechanisms for involvement of genetics providers (clinical, laboratory, educational), consumers, and others in a State/Territorial Genetics Advisory Council. The Council, together with the State Genetics Coordinator or genetics unit representative, assists in the development of the State/Territorial Plan outlined in section IC below. In states with a University Affiliated Program/Leadership Education

in Neurodevelopmental and related Disabilities (UAP/LEND) program, its representative should be involved and its activities included in the genetics advisory council and the state/territorial plan.

**C. State/Territorial Plan for Provision of Genetic Services.** The plan should include the following:

1. Assessment. A description of the:
  - a) state/territory (geography, industry, etc.)
  - b) demographic parameters (population distribution, birth rate, etc.)
  - c) state public health and genetics-related systems
  - d) system for data collection
  - e) system for evaluation of genetic services and educational activities
2. Policy Development
  - a) overview of legislative initiatives, etc.
  - b) mechanisms of funding/reimbursement of genetics services
3. Assurance
  - a) network of genetics services
  - b) system of prevention services
  - c) network of educational activities
  - d) system for periodic review of genetics services
  - e) framework of existing quality assurance (QA) measures for clinical and laboratory genetic services
4. Collaboration with all interested and relevant groups, e.g., parent support groups, sickle cell and other hemoglobinopathy programs, Muscular Dystrophy Association (MDA).

**D. Structure of the State/Territorial Genetic Services Network.** The genetic services network within a state may include several levels and types of services. (Abbreviations: MDG = MD geneticist, MSGC = MS genetic counselor, PhDMG = PhD medical geneticist)

1. Levels of Services
  - a) genetics unit of the State Health Department
  - b) large, comprehensive genetics center (public, private, academic, etc.)
  - c) genetics unit of a comprehensive managed health care facility
  - d) resident (as opposed to visiting) genetics unit within a primary health care facility (i.e., satellite or independent clinics) including MDG, MSGC, PhDMG, others
  - e) resident MSGC and/or PhDMG with periodic visits by MDG (outreach clinics)
  - f) periodic visits by MSGC, MDG, and other staff with local coordinators at outreach clinics
  - g) genetics clinics in the private sector conducted by trained MD geneticists
  - h) MSGC and/or PhDMG within single disease/medical specialty setting

- i) genetics providers within UAP/LEND programs where applicable
- j) other

## **2. Types of Services**

### **A. FAMILY FOCUSED**

- 1) state-of-the-art diagnostic, management, support, and counseling services to patients/families at all stages of the life cycle
- 2) appropriate specialty programs
- 3) evaluation

### **B. POPULATION BASED**

- 1) prenatal screening and follow-up
- 2) newborn screening and follow-up
- 3) birth defects monitoring and follow-up
- 4) teratogen information services and outcome evaluation
- 5) screening and/or evaluation in childhood and adult populations
- 6) screening and/or evaluation of selected populations, e.g., stillborns, others
- 7) educational services for professionals and the general public
- 8) data collection
- 9) evaluation

**E. Assurance.** The organization of genetic service networks must assure the following:

- 1) availability of all genetic services including comprehensive evaluation, diagnostic testing at all levels, counseling, treatment, management, and follow-up for all members of the population, irrespective of ability to pay, language differences, or education level
- 2) education of the professional and general population at all levels about important advances in genetics
- 3) development of an efficient referral system providing the population in need with the appropriate services
- 4) quality of service in compliance with accepted guidelines of laboratory and clinical services issued by organizations such as the American College of Medical Genetics (ACMG) and the College of American Pathologists (CAP).
- 5) privacy and confidentiality for patients and families
- 6) adherence to ethical and legal considerations during the provision of services (12)

**F. Funding.** Funding of clinical and laboratory genetic services should be available through:

- 1) Medicaid, Medicare
  - 2) third party carriers
  - 3) newborn screening surcharge
  - 4) state and federal service grants, e.g. Title V Block Grants, etc.
  - 5) specific disease-related organizations where applicable
  - 6) development and implementation of new CPT codes for genetics
- (13)

## II. PREVENTION

There are four levels of preventing the deleterious effects of diseases or disorders.

**A. Primary Prevention** -- the absolute prevention of the occurrence of a birth defect, genetic disorder, or disease. Feasible primary prevention measures in a public health context are as follows.

- 1) Pre- and periconceptional folic acid prophylaxis can prevent the primary occurrence of about 50% (2000) neural tube defects per year (6). Each of these infants, who might have been aborted prenatally or who, as a newborn, requires hundreds of thousands of dollars of treatment, services, and support throughout life, can now be born free of this birth defect and lead a normal life. Primary prevention includes education targeted to the relevant professionals and consumers (in this example, gynecologists, obstetricians, and women of childbearing age). Monitoring and evaluation of outcomes is an essential part of a successful primary prevention program.
- 2) Prevention of prenatal exposure to known teratogenic agents from conception through delivery can prevent the primary occurrence of deleterious effects of alcohol, cocaine, smoking, and other hazardous agents. It is estimated that 7000 infants are born each year with fetal alcohol syndrome and that tens of thousands more are in need of special education, behavioral, emotional, and learning services as a result of fetal alcohol effects (5).

Teratogen information services (TIS) play an important role in primary prevention. A TIS should be available in each state to provide information to physicians and their patients who are concerned about the risk that a particular agent will cause an unfavorable pregnancy outcome. In states without TIS, access to a national or regional information source such as Reprotox (14) and TERIS (15) should be provided. Each TIS should be a part of the national network and comply with national guidelines (16). Collaboration within the existing network has already resulted in research into the teratogenic effects of several agents and has led to the primary prevention of adverse pregnancy outcomes.

- 3) Awareness and appropriate management of maternal diseases and infections, such as PKU, diabetes, rubella, and toxoplasmosis can result in the primary prevention of birth defects and mental retardation in the newborn. Collaboration between geneticists and physicians specializing in high-risk obstetrics is essential for a successful pregnancy outcome.
- 4) Genetic counseling, which provides couples with information about their pregnancy risks and reproductive risks and pregnancy options, is another form of primary prevention. Rapid developments in alternative reproductive techniques (preimplantation diagnostics, egg donation, etc.) offer an ever-expanding set of specialized approaches to primary prevention of genetic disorders.
- 5) In the future, primary prevention will apply to those who are at risk for genetically-determined, adult-onset disorders. For example, DNA-based tests are being developed to detect those at risk for a number of different cancers. Specifically, it is estimated that current methods of screening for specific DNA mutations could detect thousands of women at risk for hereditary breast cancer. The education of professionals and families as well as the development of appropriate surveillance techniques will be essential for primary cancer prevention programs in public health.

**B. Secondary Prevention** -- Secondary prevention is the prevention of the unfavorable sequelae of already existing disorders or genotypes. Examples include the following.

- 1) Approximately 3000 affected newborns are identified each year in the U.S. through ewborn screening programs for inherited metabolic disease (8). Through detection, treatment, and follow-up, each of these infants is able to lead a life free of the deleterious consequences of their genetic disease. Newborn screening is the best paradigm for a successful genetics public health program. All 50 states and territories screen for PKU and hypothyroidism, identifying about 400 and 1200 infants respectively annually. More than 40 states also screen for sickle cell disease (>1300 cases detected) and some states screen for as many as 7 diseases. Establishing guidelines and the monitoring of newborn screening programs, through state and national committees, provide quality assurance for this highly successful operation (17, 18).
- 2) Prenatal screening offered through maternal serum markers, fetal ultrasonography, cytogenetic, and/or DNA analyses can not only identify affected fetuses and provide options about pregnancy outcome, but can identify those pregnancies in need of:
  - a) special delivery (e.g., Caesarean section to prevent damage to infants with open spine defects)
  - b) management of metabolic defects
  - c) management of prematurity or intrauterine growth retardation
  - d) prenatal intervention, including fetal surgery

- 3) Screening for genetic disorders includes:
- a) identification of those who have fragile X syndrome and other identifiable disorders
  - b) identification, at birth, of genetic disorders and other developmentally disabling birth defects requiring immediate treatment
  - c) recognizing hyperlipidemia in the population of young adults as a step toward preventing coronary artery disease
  - d) presymptomatic screening for cancers
  - e) identification of individuals at risk for adult-onset, neurodegenerative disorders

**C. Tertiary Prevention.** -- Tertiary prevention aims to ameliorate the unfavorable consequences of existing disorders. For example:

- 1) educational and other comprehensive services to children and adults with special needs
- 2) appropriate management of genetic disorders
- 3) access to orthotic and other auxiliary devices, dietary supplements, special occupational and physical therapy, ongoing support group services
- 4) referral of families to specifically oriented support groups; facilitate contact with similarly affected families.

Collaboration with consumers and the Alliance of Genetic Support Groups is of primary importance, especially in issues of tertiary prevention (19).

In addition to the three traditional levels of prevention outlined above, a 'quaternary' level of prevention involves the ongoing research into genetic diseases by the Human Genome Project, TIS, and other initiatives. Prevention also extends to the prevention of discrimination on the basis of genetic disease or testing by employers, insurers, and peers.

### **III. SERVICES**

#### **A. Types of Services**

**1. FAMILY FOCUSED SERVICES.** Genetic clinics serve children and adults at risk because of genetic disorders.

a) **General Genetic Clinics** provide service to individuals with:

- 1) known or suspected genetic disorders
- 2) congenital anomalies/birth defects

- 3) mental retardation, developmental or behavioral disorders
- 4) consanguinity or ethnicity associated with increased risk for specific disorders
- 5) family history of the above

b) **Metabolic Clinics** serve those with:

- 1) known or suspected inborn errors of metabolism regardless of onset of symptoms
- 2) a family history of a metabolic disorder
- 3) other

c) **Single Disease Clinics** are often managed by non-geneticists but require professional genetic input; these clinics serve individuals and families with:

- 1) genetic hematologic diseases (e.g., sickle cell anemia)
- 2) genetic pulmonary diseases (e.g., cystic fibrosis)
- 3) genetic neurological, neuromuscular, neurodegenerative diseases (e.g., muscular dystrophy, Huntington disease)
- 4) birth defects requiring multidisciplinary approaches to management (e.g., craniofacial disorders, spina bifida)
- 5) cancer
- 6) other genetic disorders

d) **Prenatal Clinics** focus on those at risk for an unfavorable pregnancy outcome or who have had abnormal prenatal screening results. Examples include:

- 1) risks associated with advanced maternal age
- 2) couples with a previous child affected with a genetic disorder or birth defect
- 3) couples with a family history of a genetic disorder or birth defect
- 4) couples with multiple pregnancy losses
- 5) risks associated with maternal illnesses, medications, exposures, or infections
- 6) pregnancies with abnormal screening test results
- 7) pregnancies identified as abnormal by fetal ultrasonography
- 8) other

## **2. POPULATION-ORIENTED SERVICES/SCREENING THROUGHOUT THE LIFE CYCLE**

a) **Prenatal screening:**

- 1) maternal serum alpha-fetoprotein and associated marker screening
- 2) maternal infections

- 3) maternal disease (e.g., diabetes)
- 4) carrier status (e.g., Tay-Sachs, sickle cell anemia)
- 5) fetal ultrasonography

b) **Newborn screening** and follow-up as appropriate in each state [see Newborn screening guidelines (17, 18)]

c) **Childhood screening** for genetically determined developmental disabilities, sensory deficits, and other disorders

d) **Adult screening**:

- 1) presymptomatic testing
- 2) diagnostic testing/screening
- 3) carrier testing/screening for neurodegenerative diseases in selected populations
- 4) cancer susceptibility
- 5) diseases related to aging (e.g., heart disease, Alzheimer disease)

### 3. CLINICAL LABORATORY SERVICES

Centers/clinics should have easy access to facilities for analyzing specimens for genetic studies, including, but not limited to, blood, urine, tissue, and amniotic fluid. Transport of specimens for analysis should be arranged and a tracking system maintained.

Laboratory services (13) should be provided by genetic centers and should include cytogenetic, biochemical, and molecular laboratory services as follows:

- a) Cytogenetic laboratories should be able to complete cytogenetic analysis and provide interpretation of studies of lymphocytes, amniotic fluid cells, and other tissues for the purpose of determining the number and structure of the chromosomes.
- b) Biochemical genetics laboratories should have the capacity for analysis and interpretation of test results for alpha-fetoprotein, selected enzymes, substrates, metabolic reactions, structural proteins, hemoglobins, and other biochemical systems.
- c) Molecular genetics laboratories should have the capacity to complete specified DNA analysis and interpret results.
- d) Requests for biochemical and molecular tests which are unavailable in a state should be referred to regional or national facilities as needed.

Effective communication between patients, clinics, laboratories and primary care physicians must be ensured. Accurate and timely interpretation of laboratory results with supportive explanation, counseling, follow-up, and referrals must be available.

**B. Levels of Service.** See section I. Organization and Administration, paragraph D.

### **C. General Facility and Operational Requirements**

#### **1. GENERAL FACILITY REQUIREMENTS**

- a) The facility should be an identifiable unit in an accredited state or other medical school, a hospital, or a clinic accredited by the Joint Commission on Accreditation of Health Care Organizations.
- b) The facility should be licensed by the State Department of Health, if such licensure is required for operation, or by any other licensing agency as required.
- c) Private facilities should demonstrate compliance with appropriate certifying agencies.
- d) The facility should have access to medical support services necessary for diagnosis of genetic or congenital disorders.
- e) The facility should include, but not be limited to, an identifiable clinic area with rooms for examination, counseling, management, and evaluation which are appropriately equipped for delivery of services and privacy for patients and/or family.
- f) The facility must be accessible to the handicapped.
- g) The facility should have its own telephone number or extension through which all services can be accessed.

#### **2. GENERAL OPERATIONAL REQUIREMENTS FOR SERVICES**

- a) Services should be available, accessible and culturally appropriate.
- b) Admission and referral policies should facilitate entry of the population to be served.
- c) The administration and staff of the center should continuously update their knowledge and skills through in-service education programs and attendance at conferences, seminars, and workshops.
- d) The center should develop and maintain an active program to monitor the quality of services provided.

e) Input by those using the services ('consumer input') should be routinely obtained for the purpose of planning evaluation of services.

f) Laboratories associated with the genetics unit should participate successfully in available proficiency testing programs.

The centers should ensure effective and efficient administration.

g) Staff meetings should be scheduled at least quarterly with written meeting summaries maintained.

h) The center should maintain written contracts/agreements for all core professional services not directly provided by personnel of the center. Contracts or agreements should include identification of services to be rendered, including, where appropriate, the hours and personnel involved as well as the payment and billing procedures.

i) Centers should maintain written protocols identifying laboratories that will accept specimens for necessary tests, payment methods, unique services, and typical turnaround time used for diagnostic evaluations. A tracking system (log) should be maintained for all specimens.

j) A written admission policy should be available and include: service fees, billing procedures, and available financial assistance, as well as schedulers of clinic and office hours.

k) No individual with a suspected genetic condition should be refused genetic services because of any disability or medical condition.

l) State programs should provide support to those patients/families who are unable to pay.

## **D. Genetic Health Care Professionals**

### **1. STAFF**

The following staff should be available or accessible to provide genetic services at each center. Alternatively, expert consultation must be available by referral to another institution. Genetic centers typically have as their director a medical geneticist who heads a staff consisting of one or more of the following genetic health care professionals.

*In this section and in the corresponding sections in Appendix C, superscript numbers are used to explain the sources of the wording:*

<sup>1</sup>*The wording for categories a,b,d,e and f is taken verbatim from the American Board of Medical Genetics (ABMG) Bulletin of Information, 1996 (20).*

<sup>2</sup>*Category c is from the American Board of Genetic Counseling (ABGC) Bulletin of Information, 1996 (21).*

<sup>3</sup>*Categories h and i are from International Society of Nurses in Genetics (ISONG) Standard of Practice, DRAFT, 1996 (22).*

<sup>4</sup>*Paragraphs are abstracted from The ABMG Bulletin of Information, 1996 (20).*

<sup>5</sup>*Paragraphs are abstracted from document ABGC Bulletin of Information, 1996 (21).*

<sup>6</sup>*Association of Cytogenetic Technologists (ACT) (23).*

- a) Clinical Geneticist<sup>1</sup> An individual who holds an MD or DO degree and demonstrates competence to provide comprehensive diagnostic, management, and counseling services. Clinical geneticists come from a variety of disciplines including pediatrics, internal medicine, obstetrics/gynecology, ophthalmology, and dentistry.<sup>4</sup>
- b) PhD Medical Geneticist<sup>1</sup> An individual with a PhD degree who works in association with a medical specialist, is affiliated with a clinical genetics program, serves as a consultant to medical and dental specialists, and/or serves in a supervisory capacity in a medical genetics program. PhD Medical Geneticists have PhD's in a variety of disciplines including biochemistry, molecular biology, epidemiology, and mathematics.<sup>4</sup>
- c) Genetic Counselors<sup>2</sup> Genetic Counselors are health professionals with a Master's degree who are academically and clinically prepared to provide genetic counseling services to individuals and families seeking counseling information about the occurrence, or risk of recurrence, of a genetic condition or birth defect. They are prepared to practice as an integral part of a genetic services delivery team. Genetic Counselors come from a variety of backgrounds including biology and other basic sciences, social work, and nursing.<sup>5</sup>
- d) Clinical Cytogeneticist<sup>1</sup> An individual with a doctoral degree (MD, DO, PhD) who is competent to perform and interpret cytogenetic analyses relevant to the diagnosis and management of human genetic disease and can act as a consultant regarding laboratory diagnosis for a broad range of disorders.
- e) Clinical Biochemical Geneticist<sup>1</sup> An individual with a doctoral degree (MD, DO, PhD) who is competent to perform and interpret biochemical analyses relevant to the diagnosis and management of human genetic disease, and who acts as a consultant regarding laboratory diagnosis of a broad range of disorders.

- f) Clinical Molecular Geneticist<sup>1</sup> An individual with a doctoral degree (MD, DO, PhD) who is competent to perform and integrate molecular analyses relevant to the diagnosis and management of human genetic disease, and who acts as a consultant regarding laboratory diagnosis of a broad range of disorders.
- g) Cytogenetic Technologist<sup>6</sup> An individual with a minimum of a BS degree who demonstrates competence to provide cytogenetic analysis in a clinical diagnostic laboratory under the supervision of a laboratory director qualified in clinical cytogenetics.
- h) Genetic Nurse<sup>3</sup> An individual who provides nursing care for a client population with a specific genetic condition or a need for a specific genetic service. Genetic Nurses are licensed registered nurses who have received genetic continuing education.
- i) Advance Practice Nurse in Genetics<sup>3</sup> An individual with a MS or PhD in nursing who has completed graduate level genetics course work and assures possession of current knowledge through participation in genetic continuing education.
- j) Perinatologist/Obstetrician or other physician must be accessible for referral; conducts all invasive prenatal diagnostic studies.
- k) Other medical/surgical specialties and subspecialties available through a clinical genetic center should include, but not be limited to: pediatrics, obstetrics and gynecology, pathology, psychiatry, neurology, and orthopedics.
- l) Other staff available through a clinical genetics center should include, but not be limited to: psychologist, social worker, nutritionist, occupational and physical therapists, special education experts, foreign language translators, and interpreters for the hearing impaired.

## **2. STAFF CREDENTIALS**

- a) The following providers of genetic services: Clinical Geneticists; PhD Medical Geneticists; Genetic Counselors; Cytogenetic, Biochemical, and Molecular Genetic Laboratory Directors; and Cytogenetic Technologists should be certified, as appropriate, by the American Board of Medical Genetic (ABMG), the American Board of Genetic Counseling (ABGC), the National Certification Agency for Medical Laboratory Personnel (NCA), or be board eligible. Eligible status for certification may be maintained for no more than two administrations of the Board's examination.
- b) Physicians should be licensed by the state and be board certified or board eligible in their speciality area.

- c) A Clinical Geneticist should be a licensed physician.
- d) Medical directors should be licensed physicians.
- e) Cytogenetic Technologists should be certified by NCA and licensed by the state as appropriate.
- f) All other professionals should be licensed by the state, as appropriate.
- g) If no license requirements exist, the professional should be certified (accredited) by the appropriate national organization.

**E. Components of a Genetic Evaluation.** The following clinical genetic services should be provided, on site, unless otherwise specified, by appropriately credentialed professionals.

**1. GENERAL CLINICAL GENETIC SERVICES** should include, as appropriate:

- a) obtain family history/construct pedigree
- b) obtain personal medical history, review patient and family medical records
- c) physical examination including growth and development assessment
- d) access to diagnostic testing including, but not limited to, radiological procedures (X-rays, magnetic resonance imaging [MRI])
- e) evaluation and diagnosis
- f) specimen collection for diagnostic studies/evaluations (inpatient and outpatient)
- g) risk assessment
- h) genetic counseling and education including anticipatory guidance for patient/family, support
- i) management/treatment of genetic diseases or conditions, including dietary and nutrition counseling as relevant and appropriate
- j) coordination of medical/surgical consultation(s) and/or referral(s) for supporting services as appropriate
- k) short- or long-term follow-up as needed
- l) staff should be familiar with practice guidelines developed for specific disorders or groups of disorders by the American College of Medical Genetics subcommittee on practice guidelines. Examples include the dysmorphic newborn, the developmentally delayed child, stillborns, and individuals or families with breast cancer (2 and Appendix E).
- m) national guidelines for presymptomatic testing should be consulted/observed for relevant disorders, e.g., Huntington, familial cancers, etc (24).

**2. PRENATAL GENETIC SERVICES** for problems relating to increased risk should, as appropriate, include:

- a) review of medical records and history

- b) analysis of family history/pedigree construction
- c) physical examination
- d) other screening and/or diagnostic procedures, including but not limited to, cytogenetic, biochemical, and molecular testing for patient/family
- e) risk assessment
- f) genetic counseling, education, and support for patient and family
- g) management/treatment of genetic diseases or conditions
- h) coordination of medical consultation(s) and/or referral(s) for services, as appropriate
- i) The center should provide or arrange for radiological or other diagnostic testing, including but not limited to, high resolution ultrasonography, fetal echocardiography, amniocentesis, chorionic villus sampling, tissue biopsy, X-ray, magnetic resonance imaging (MRI), CT scan, and cordocentesis.
- j) follow-up, communication of results, further consultation as needed
- k) staff familiarity with Practice Guidelines (see section E.1.1)

**3. PRIOR TO ANY TESTING PROCEDURE**, every person seeking services will be advised of:

- a) the nature and purpose of the procedure and its implications
- b) benefits and risks involved
- c) the opportunity to decline participation
- d) estimated fees, charges, and billing procedures

**4. A WRITTEN SUMMARY/LETTER** of the results of a genetics evaluation and its implications should be sent to the referring physician(s). A copy should be placed in the patient's chart and, where possible, the results sent in the form of an understandable letter to the patient/family. The opportunity to discuss all issues with a supportive, informed professional should be offered. Relevant literature and support educational materials should be available.

**5. APPROPRIATE REFERRALS** should be made to the following:

- a) medical specialties/subspecialties
- b) care management services
- c) social services
- d) early intervention services
- e) home health services
- f) national/local family support groups (19)

## **F. Patient Records**

1. Confidentiality of records must be protected and written procedures regarding access to records must be known by all staff.

2. It must be standard operating procedure to obtain necessary releases and send a written report on each patient and/or family to the referring physician/professional.
3. Prior to releasing a patient's report to any other professional service provider, a specific written release must be signed by the patient/parent.
4. Genetic records should be maintained as part of the permanent medical record for each patient. Records should be retained in confidential files which are locked or otherwise secured. Records should be accessible to staff of the center and consultants and should include, but not be limited to:

- intake information
- medical history
- laboratory test results
- diagnostic reports
- counseling summary report
- plan of care (as indicated)
- record of services at other facilities
- informed consent forms
- written releases
- referral information

### **G. Human and Legal Rights**

1. There must be a written informed consent policy for all evaluations as appropriate and for treatment procedures.
2. Services must be culturally appropriate and competent. The center should arrange for foreign language interpreters and interpreters for the hearing impaired when necessary.
3. The center should not discriminate through admission policies, hiring policies, or promotional opportunities on the basis of race, religion, ethnic origin, sex, or handicapping condition.
4. Institutional review board guidelines must be observed when conducting research.
5. Patients/families must be informed about ownership and fate of stored biological specimens and all relevant issues related to such specimens.
6. Patients and family should have access to information about the hospital's or institution's bio-ethics committee and its deliberations relevant to the patient/family's specific situation as applicable.
7. See also item m) under section E.1. General Clinical Genetic Services.

## **H. Quality Assurance**

1. The administration and staff of the center must demonstrate a commitment to quality care:

- a) There should be a written statement of mission and goals for the center.
- b) There should be a designated administrator for the service unit.
- c) The administration and staff of the center should continuously update their knowledge and skills through in-service educational programs and attendance at conferences, seminars, and workshops.
- d) The center should develop and maintain an active program to monitor the quality of services provided.
- e) Consumer input (e.g., patient satisfaction) should be routinely obtained for the purpose of planning and evaluation of services.
- f) Laboratories associated with the genetic unit shall participate successfully in the appropriate proficiency testing programs.

## **IV. RESEARCH<sup>1</sup>**

A. The state genetics unit and/or the genetics coordinator together with the advisory council, should be aware of educational sessions, media releases or other vehicles, whereby results from research are communicated accurately and appropriately to those professionals and the general public for whom they are relevant.

B. Patients and families should be given the option to participate in research studies if desired under the following conditions:

- 1) All research projects/protocols involving human subjects have undergone review and obtained approval of the Institutional Review Board.
- 2) Patients/families are fully aware that they are participating in research and most likely will receive no answers or results from their tests.
- 3) Patients/families have given informed consent after receiving an accurate and understandable explanation of the procedure in question, its risks, benefits, and other implications.

<sup>1</sup>Research training for genetics professionals is not considered to be relevant to this document

4) Privacy/confidentiality of the patient's participation and results are ensured.

C. State genetics units should be encouraged to participate in collaborative, public health-related research projects, e.g., TIS-initiated research, epidemiologic data collection, etc.

**V. EDUCATION.** In the broadest sense, education is provided at several levels of sophistication to different target audiences. Levels of education include:

**A. AWARENESS** - i.e., a "flash" of information about the existence of a subject. For example:

- 1) BRCA1 is a recently recognized gene which might in the future enable the detection of some individuals at risk for developing breast cancer.
- 2) Pregnancies which are exposed to hazardous environmental agents might be at higher risk for an unfavorable outcome.
- 3) Disorders which occur within several members of a family might be hereditary.
- 4) Taking folic acid prior to and during pregnancy can prevent some birth defects.

"Awareness increasing" activities might include TV spots, flyers, news conferences, notices/posters in public (or clinical) locations, individual items mentioned within educational sessions.

Target audiences for awareness are broad and include the general public (all ages), teachers, media personnel, school children, nongenetics professionals, others. The public health agencies within states can take responsibility for increasing the awareness among professionals and the public when important issues arise. It is they who produce flyers, posters, and TV flashes.

**B. INFORMATION** - Information implies a more detailed level of knowledge than does awareness.

For example:

- 1) BRCA1 may be a tumor suppressor gene which, if mutated, might predispose to breast and/or ovarian cancer.
- 2) If a physician, patient, or other care giver is concerned about hazards to a pregnancy, teratogen information services can provide more information regarding a particular agent.
- 3) There are multiple mechanisms and patterns of inheritance; for example, not all familial disorders are genetic.

4) Which birth defects might be prevented by folic acid ingestion, what dose is recommended, and to whom does this information apply.

Target audiences for information include teachers at all levels: grades 6-12, college; topic oriented non-genetics professionals, medical students, and other trainees; consumers; relevant parents/families; media personnel; and specific interest groups.

Public health agencies support workshops, produce brochures, etc. and are generally involved with the information component of education also.

**C. INSTRUCTION** - Instruction implies providing more complicated information such as:

1) the state of the art of BRCA1 screening, including the molecular structure of the gene and how it functions to cause cancer, who should be offered screening, what results can be expected, and how are they interpreted.

2) mechanisms of the effects of alcohol exposure during pregnancy

Instruction occurs during single topic educational sessions, workshops, "hands-on" discussions, and CME conferences. Target audiences for instruction are usually those for whom the subject is personally or professionally relevant. These audiences include genetics and non-genetics service providers, MD specialists, teachers, students, allied health professionals, creators of media teaching tools, specific at-risk populations, support groups, parents, patients, and others.

Evaluation should be a required component of each level of education including awareness (less important), informational and instructional activity (very important).

**D. TRAINING** - is a long term activity, the anticipated result of which is the emergence of individual experts within a specific area. It includes training of a) future genetics professionals at all levels and in all areas of expertise, and b) existing genetics professionals in new areas of their field. In the current climate of managed care, it is recommended that MDs in other areas of expertise become acquainted (e.g., through short term fellowships, independent study courses, collaboration, training sessions, etc.) with the basic principles of modern genetics. It is recommended that this occur principally with primary care physicians. Other care givers within specific settings also require additional training to become experts in their own area of activity.

Participants in training programs are eligible for accreditation and certification by national regulatory bodies, e.g., ABMG, RRC, ABGC, NCA, etc. State public health agencies have no known role in training of genetics professionals, unless it is formal, subject specific training.

MCH training programs in schools of public health should be encouraged to ensure that public health students become aware of the existence of genetic services and of the role of genetics and prevention in a comprehensive health care system. For example, trained professionals are needed in e.g., epidemiology, data collection and follow up upon which plans for genetic testing, screening, reporting can be based. It is recommended that genetics courses be offered in schools of public health.

## **VI. DOCUMENTATION OF NEEDS AND SERVICES**

### **A. DATA SOURCES**

At the state level, numerous public health databases collect information concerning individuals with genetic diseases. These data systems are generally administrative in nature, providing documentation of vital status, eligibility for and utilization of program services, and, in some instances, monitoring or surveillance functions. For assessment of genetics in public health, the purpose and primary functions of each database must be considered in the initial assessment of its utility for this purpose. For example, birth defects registries are now in existence in about half of the states. These registries vary greatly in design, case ascertainment methods, and primary uses.

A partial list of potential statewide data sources for genetics and public health would include:

1. state level clinical genetics databases (at a minimum, the CORN, or Regional, Minimum Data Set) (25)
2. newborn screening data
3. vital statistics: birth, fetal death, and death certificates
4. statewide hospital discharge data (linked at the individual level across in-patient stays)
5. Medicaid/Medicare eligibility, claims, and provider datasets
6. statewide/local cytogenetics registry data
7. statewide birth defects registry (a few areas have local registries)
8. statewide/local population based cancer/tumor registries or reporting systems
9. other registries (developmental disabilities surveillance, support group registries, specialized support group registries)
10. directories of genetics service providers and referral sources

11. cytogenetic laboratory databases collected by the ACT
12. federal census data (primarily for population denominators and as basis for population projections)
13. Special surveys and research projects include:
  - a) Pregnancy Risk Assessment Monitoring System (Centers for Disease Control and Prevention (CDC) - ongoing, many but not all states participate)
  - b) National Maternal and Infant Health Survey (National Center for Health Statistics (NCHS) - most recent is 1988)
  - c) Behavioral Risk Factor Surveillance System (CDC - annual, almost all states participate)
  - d) National Survey of Family Growth (NCHS)

## **B. LINKAGES**

There are a number of essential linkages which should occur at every state level for the purpose of monitoring occurrence of specific genetic diseases, outcomes in infants/children with those diseases, and assessment of service utilization and efficiency of service delivery. These include:

- 1) linkage of birth and death certificates for all deaths up to age six
- 2) linkage of birth defects and tumor registry data for all pediatric cancer cases
- 3) routine linkage of birth defects registry records with vital statistics (births, fetal deaths and deaths)
- 4) routine linkage of statewide inpatient hospital discharge records with birth certificates
- 5) routine linkage of newborn screening records with birth certificates
- 6) linkage of (MSAFP/AFAFP/triple screen, etc.) screening database with vital statistics (if such a database exists)
- 7) linkage, at least in the form of numerator/denominator ratio data, between the statewide clinical genetics services database and birth/fetal death certificates
- 8) systems for direct referral from clinical genetics to early intervention services for infants and children under the age of 3 (Part H: Public Law 99-457), Children with Special Health Care Needs (CSHCN), Supplemental Social Insurance (SSI) and other

services/entitlements for children/families with disabilities associated with genetic disorders or diseases.

## **VII. FUNDING**

All State Public Health Agencies are responsible for funding several aspects of genetics services. However, the mechanisms, organization and types of funding differ among states, hence details cannot be presented. Types of funding in most states include:

- 1) Medicaid, Medicare
- 2) third party carriers, including employers/insurers
- 3) newborn screening surcharge
- 4) state and federal grants, e.g. Title V, MCH Block Grants, others
- 5) specific disease or disease group organizations, e.g., Muscular Dystrophy Association, Cystic Fibrosis Foundation
- 6) specific sources for individuals with developmental disabilities, e.g., Bureau for Children with Special Health Care Needs, "Crippled" Children's organizations, etc.

New CPT codes for genetic services are being developed which will result in improved reimbursement for genetics (13). In Texas specific genetic (G)CPT codes have been developed and are currently in use.

Third party payment for genetic services is often a problem. There are a considerable number of patients in need of and/or receiving genetics services who a) have no insurance coverage; or b) do not wish to inform the insurance carrier about the disorder for fear of discrimination. It is important that HMOs continue to be informed about the benefits of genetic services and about the implications for the of some of the new genetic technology.



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## APPENDICES

- A. CORN AND THE REGIONAL NETWORKS
- B. CALCULATIONS
- C. DESCRIPTIONS OF GENETIC PROFESSIONALS
- D. DEFINITIONS OF SALIENT TERMS
- E. LIST OF PUBLISHED STATEMENTS ON HEALTH SUPERVISION FOR CHILDREN WITH GENETIC DISORDERS, DEVELOPED BY AMERICAN ACADEMY OF PEDIATRICS, COMMITTEE ON GENETICS



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## APPENDIX B

## CALCULATIONS

B-1. ~4,000,000 live births/year

Incidence of NTD = 1/1000

50% preventable by folic acid prophylaxis

50% of 4000 = 2000

If only \$10,000 cost/affected infant, then  $2000 \times \$10,000 = \$20,000,000$

B-2. 6% of 4 million pregnancies = 240,000

Incidence of nondisjunction at age 35-45 u 2%

2% of 240,000 = 5000 infants



## APPENDIX C

### DESCRIPTIONS OF GENETIC PROFESSIONALS\*

#### CLINICAL GENETICIST:

An individual who holds an MD or DO degree and demonstrates competence to provide comprehensive diagnostic, management, and counseling services.

Competence in this area implies that the individual possesses:

- diagnostic and therapeutic skills in a wide range of genetic disorders;
- an appreciation of the heterogeneity, variability, and natural history of genetic disorders;
- the ability to elicit and interpret individual and family histories;
- the ability to integrate clinical and genetic information and appreciate the limitations, interpretation, and significance of specialized laboratory and clinical procedures;
- the expertise in genetic and mathematical principles to perform risk assessment;
- the skills in interviewing and counseling techniques required to: 1) elicit from the patient or family the information necessary to reach an appropriate conclusion; 2) anticipate areas of difficulty and conflict; 3) help families and individuals recognize and cope with their emotional and psychological needs; 4) recognize those situations requiring psychiatric referral; and 5) transmit pertinent information effectively (i.e. in a way that is meaningful to the individual or family);
- the knowledge of available health care resources required for appropriate referral.

Clinical Geneticists come from a variety of disciplines including pediatrics, internal medicine, obstetrics/gynecology, ophthalmology, and dentistry. Certification is provided through the American Board of Medical Genetics.

#### Ph.D. MEDICAL GENETICIST:

An individual with a Ph.D. degree who works in association with a medical specialist, is affiliated with a clinical genetics program, serves as a consultant to medical and dental specialists, and/or serves in a supervisory capacity in a medical genetics program.

Competence in this area implies that the individual possesses:

- the ability to elicit and interpret individual and family histories;
- an appreciation of the heterogeneity, variability, and natural history of genetic disorders;

\*This document has been abstracted from language approved by the American Board of Medical Genetics, the American Board of Genetic Counseling, and the Association of Cytogenetic Technologists.

- the ability to integrate clinical and genetic information in order to appreciate the limitations, interpretation, and significance of specialized laboratory and clinical procedures;
- the expertise in genetic and mathematical principles to perform complex risk assessments, to interpret pedigree analysis (both segregation and linkage) and to understand the principles of genetic etiology;
- the skills in interviewing and counseling techniques required to: 1) elicit from the patient or family the information necessary to reach an appropriate conclusion; 2) anticipate areas of difficulty and conflict; 3) help families and individuals recognize and cope with their emotional and psychological needs; 4) recognize those situations requiring psychiatric referral; and 5) transmit pertinent information effectively (i.e. in a way that is meaningful to the individual or family).

Ph.D. Medical Geneticists have Ph.D.'s in a variety of disciplines including biochemistry, molecular biology, epidemiology, and mathematics. Certification is provided through the American Board of Medical Genetics.

### **CLINICAL CYTOGENETICIST:**

An individual with a doctoral degree (M.D., D.O., Ph.D.) who is competent to perform and interpret cytogenetic analyses relevant to the diagnosis and management of human genetic diseases and can act as a consultant regarding laboratory diagnosis for a broad range of disorders.

Competence in this area implies that the individual possesses:

- the ability to supervise and direct the operations of clinical cytogeneticists diagnostic laboratory;
- an appreciation of the heterogeneity, variability, and natural history of genetic disorders;
- diagnostic and interpretive skills in a wide range of cytogenetic problems;
- the ability to appropriately communicate cytogenetic laboratory results in the capacity of consultant to other clinicians or directly to patients in concert with other appropriate clinicians or genetic counselors.

Clinical Cytogeneticists generally have a Ph.D. in molecular biology, molecular genetics, or cytogenetics. Certification is provided through the American Board of Medical Genetics.

### **CYTOGENETIC TECHNOLOGIST**

An individual with a minimum of a BS degree who demonstrates competence to provide cytogenetic analysis in a clinical diagnostic laboratory under the supervision of a laboratory director qualified in clinical cytogenetics.

Competence in this area implies that the individual possesses:

- the ability to process specimens for cytogenetic analysis, including the knowledge to select culture, harvesting, slide preparation, and staining techniques appropriate to each specimen type;
- the skill to: 1) select the appropriate metaphases, identify chromosomal abnormalities, assess difficulties with analysis, and prepare accurate karyotypes; and 2) summarize the results and prepare reports which are reviewed by the laboratory director or another clinical cytogeneticist;
- knowledge of general laboratory skills, quality control and quality assurance procedures, and knowledge of the general principles of biology and genetics, including the principles of clinical cytogenetics.

Cytogenetic Technologists come from a variety of backgrounds and include biologists, chemists, and clinical laboratory scientists. Certification as a Clinical Laboratory Specialist in Cytogenetics is provided through the National Certification Agency for Medical Laboratory Personnel (NCA) and is maintained through participation in continuing education.

### **CLINICAL BIOCHEMICAL GENETICIST**

An individual with a doctoral degree (M.D., D.O., Ph.D.) who is competent to perform and interpret biochemical analyses relevant to the diagnosis and management of human genetic disease, and who acts as a consultant regarding laboratory diagnosis of a broad range of disorders.

Competence in this area implies that the individual possesses:

- the ability to supervise and direct the operations of a clinical biochemical diagnostic laboratory;
- broad knowledge of: 1) basic biochemistry and biology; 2) the application of biochemical techniques to the diagnosis and management of genetic diseases; and 3) the etiology, pathogenesis, clinical manifestations, and management of human inherited biochemical disorders;
- the ability to appropriately interpret and communicate biochemical laboratory results in the capacity of consultant to other clinicians or directly to patients in concert with other appropriate clinicians or genetic counselors.

### **CLINICAL MOLECULAR GENETICIST:**

An individual with a doctoral degree (M.D., D.O., Ph.D.) who is competent to perform and integrate molecular analyses relevant to the diagnosis and management of human genetic diseases, and who acts as a consultant regarding laboratory diagnosis of a broad range of disorders.

Competence in this field implies that the individual possesses:

- the ability to supervise and direct the operations of a clinical molecular genetics diagnostic laboratory;
- the ability to perform a variety of diagnostic assays;
- a broad knowledge of: 1) basic molecular biology and genetics; 2) the application of recombinant DNA techniques and linkage analysis to the diagnosis of genetic diseases; and 3) the etiology, pathogenesis, clinical manifestations, and management of human genetic disorders;
- the ability to appropriately interpret and communicate molecular diagnostic laboratory results in the capacity of a consultant to other clinicians or directly to patients in concert with other clinicians or genetic counselors.

Clinical Molecular Geneticists come from a variety of backgrounds and include medical geneticists and individuals with a Ph.D. in molecular genetics or molecular biology. Certification is provided through the American Board of Medical Genetics.

### **GENETIC COUNSELOR:**

Genetic Counselors are health professionals with a Master's degree who are academically and clinically prepared to provide genetic counseling services to individuals and families seeking counseling information about the occurrence, or risk of recurrence, of a genetic condition or birth defect. They are prepared to practice as an integral part of a genetic services delivery team.

Competence in the area of genetic counseling implies that the individual possesses the ability to:

- elicit and interpret individual, family, medical, developmental, and reproductive histories;
- determine the mode of inheritance and risk of transmission of genetic conditions and birth defects;
- discuss the mode of inheritance, features, natural history, means of diagnosis, and management of these conditions;
- identify, coordinate, interpret, and explain genetic laboratory tests and other diagnostic studies;
- assess psychosocial factors, recognizing social, educational, and cultural issues;
- evaluate the client's/family's responses to the condition or risk of recurrence and provide client-centered counseling and anticipatory guidance;
- communicate information to family members in an understandable manner;
- facilitate informed decision making about testing, management, and reproductive alternatives; identify and effectively utilize community resources that provide medical, educational, financial, and psychosocial support and advocacy; and provide accurate written documentation of medical, genetic, and counseling information for families and health care professionals.

Genetic Counselors come from a variety of backgrounds including biology and other basic sciences, social work, and nursing. Genetic Counselors are certified through the American Board of Genetic Counselors.

### **ADVANCED PRACTICE NURSE IN GENETICS**

An individual with a M.S. or Ph.D. in nursing who has completed graduate level genetics course work and assures possession of current knowledge through participation in genetic continuing education.

Competence in this area implies that an individual possesses the ability to utilize the nursing process in practice delivery as listed under genetic nurse as well as have the ability to:

- use counseling skills and interventions to assist clients in understanding genetic concepts, their implications to the client and family, and assist the client in adjusting to their perceived burden;
- provide consultation to health care providers and others to influence the plan of care and enhance the abilities of others to provide care for patients with genetic conditions;
- participate in the clinical evaluation of clients with genetic conditions;
- guide nurses in the specialized care of client's with genetic conditions; provide expert input into the development, management, and/or evaluation of a multi disciplinary genetic clinical research protocol;
- participate in assessment and deliberation of ethical, legal, and social consequences of existing and predicted genetic services and technologies;
- provide case management across a variety of settings for genetic clients who have complex health care needs.

Advanced Practice Nurses in genetics come from a variety of nursing disciplines including maternal/child health, oncology, neurology, hematology, endocrine, and others. Steps towards creating a nursing certification examination in clinical genetics are in process. Some genetics Advanced Practice Nurses have become certified as genetic counselors through the American Board of Genetic Counseling.

### **GENETIC NURSE:**

An individual who provides nursing care for a client population with a specific genetic condition or a need for a specific genetic service. Genetic Nurses are licensed registered nurses who have received additional education in the area of genetics.

Competence in this area implies that the individual possesses the ability to:

- collect and examine health data by participating in activities such as performing a physical examination; obtain family, medical, developmental and reproductive histories; collect appropriate laboratory data; inquire into client's desired health outcomes; and assess the client's understanding of the genetic condition;
- establish an appropriate plan of nursing care designed for the genetic client and coordinate that care with other health professionals. Client focused immediate and long term health care needs are determined and used to develop a plan of action;
- implement interventions which may include: 1) heightening awareness about services and health behavior that may reduce the risk of or symptoms of a genetic condition; 2) facilitate successful adaptive responses to disease processes; 3) educate about, administer, and monitor responses to therapies for a genetic condition; 4) advocate for and facilitate access to genetic resources and support groups; and 5) provide or reinforce information about a genetic condition routinely cared for by the nurse;
- evaluate the plan of care based on new data, resources, and the client's changing needs

Genetic Nurses can be found in a diverse number of clinical settings specific to the disorder in question.

## APPENDIX D

### DEFINITIONS

**Birth Defect** - An abnormality of structure or function present at birth irrespective of its cause.

**Congenital** - Anything that is present at birth, e.g., a missing finger at birth is a congenital abnormality (irrespective of its cause) whereas a finger amputated in an accident is an acquired abnormality.

**Genetic Counseling** - A communication process which deals with the human problems associated with the occurrence, recurrence or the risk thereof, of a genetic disorder within a family. This process involves an attempt by one or more appropriately trained persons to help the individual or family: (1) comprehend the medical facts, including the diagnosis, the probable course of the disorder, and the available management; (2) appreciate the way heredity contributes to the disorder, and the risk of recurrence in specified relatives; (3) understand the options for dealing with the risk of recurrence; (4) choose the course of action which seems most appropriate to them in view of their risk and the family goals and act in accordance with that decision; and (5) make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder. Reassurance that an individual is NOT at risk for the occurrence or recurrence of a disorder is also an inseparable component of genetic counseling.

Genetic counseling is characteristically performed by a master's level trained, board certified genetic counselor and is usually preceded by a genetic evaluation of the patient/family.

**Genetic Diagnosis/Evaluation** - The process of determining the presence or absence of a condition through physical evaluation and/or laboratory testing. Once a diagnosis has been made, appropriate genetic counseling can be performed and an acceptable treatment plan developed. Diagnostic procedures performed that focus on a fetus are referred to as prenatal services while all others are referred to as clinical services. The clinical evaluation and diagnostic process is typically performed by a physician who is certified by the American Board of Medical Genetics as a clinical geneticist, while prenatal evaluation and diagnostic testing may be performed by an obstetrician/perinatologist, in consultation with genetic professionals as needed.

**Genetic Disorders** - Disorders associated with a change in the genetic material, i.e., in the chromosomes, the genes or the DNA, with or without a contribution of environmental factors.

**Genetic Screening** - A process by which an individual undergoes a procedure and/or test to indicate if they are at a greater or lesser risk than the general population for having a specific condition. If a positive screening result is encountered, the individual is typically referred for diagnostic (confirmatory) testing. Current examples of genetic screening include newborn

screening for PKU, or maternal serum alpha-fetoprotein screening during pregnancy to identify fetuses with spina bifida or Down syndrome.

**Genetics** - The branch of biology which deals with heredity and variation.

Human Genetics - The branch of biology which deals with heredity and variation in the human species.

Medical Genetics - The application of human genetics to diseases and abnormalities of human development.

Clinical Genetics - The provision of medical services to individuals, families and populations who have or are at risk for disorders with genetic implications.

## APPENDIX E

### PRACTICE GUIDELINES Developed by the Committee on Genetics of the American Academy of Pediatrics

The Committee has now completed statements on health supervision for children with the following genetic disorders. They are published in Pediatrics:

- Down syndrome      Pediatrics              93:855-859, 1994
- Achondroplasia      Pediatrics              95:443-451, 1995
- Neurofibromatosis   Pediatrics              96:368-372, 1995
- Turner syndrome    Pediatrics              96:1166-1178, 1995
- Fragile X              Pediatrics              98:297-300, 1996
- Sickle cell disease   Pediatrics              98:467-472, 1996
- Marfan syndrome    Pediatrics              (to be published November 1996)
- Newborn screening   Pediatrics              98:473-501, 1996

fact sheets

The Committee also discussed plans to develop a compendium for pediatricians and primary care physicians which would include the afore cited health supervision statements as well as an introduction discussing the approach to the infant and child with a congenital malformation, a genetic assessment form (currently in development...suggest best form to be reviewed by the Section and the College of Medical Genetics), as well as previously published Committee statements (e.g., issues in Nb screening, maternal serum-AFP testing, congenital hypothyroidism and prenatal genetic diagnosis for pediatricians).

The Committee is also preparing for publication a summary of a recent workshop on phenylketonuria and an update related to issues in the management of patients with phenylketonuria.