Interagency Council for Genetic Services

Resource Allocation Plan
2008-2009

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Interagency Council for Genetic Services
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2008-2009 Resource Allocation Plan

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PREFACE

The Interagency Council for Genetic Services (IACGS) was established in 1987 (70(R)) to survey current resources for human genetic services in the state, evaluate current and future needs for services, assist in coordinating statewide human genetic services, and monitor the provision of human genetic services.

Members include one representative each from:

- The Texas Department of State Health Services (DSHS);
- The Texas Department of Aging and Disability Services (DADS);
- The Texas Department of Insurance (TDI);
- The University of Texas Health Science Centers (UTHSC);
- The providers that contract with DSHS to provide genetic services; and

Two representatives who are:

- Consumers of genetic services, or representatives of consumer groups related to the provision of genetic services.

Human Resources Code Title 9, Chapter 134, Section 134.0041 directs the Council to biennially develop a resource allocation plan recommending how funds for genetic services should be spent during the next fiscal biennium. The report inventories available resources, identifies gaps and barriers to service, discusses challenges and opportunities, and recommends needed action to assure access to quality care.

Questions regarding the 2008-2009 Resource Allocation Plan may be directed to:

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EXECUTIVE SUMMARY

Rapid technological advances in the field of genetics have led to a better understanding of the genetic basis of disease. Genetic disorders traditionally have been associated with problems of pregnancy and birth. We now understand that genetics plays a role in the development of many common diseases, many of which do not appear until later in life. This has implications for the provision of genetic services across the lifespan and the integration of genetic knowledge across medical specialties.

Information gleaned from the Human Genome Project and subsequent biomedical research has led to unprecedented breakthroughs in the diagnosis and management of disease, including increased genetic testing capabilities, and the availability of individually tailored treatment. With these advances come challenges surrounding the issues of readiness and capacity. Can our current genetics workforce meet increasing demands from the public for genetic advice? How do we keep pace with the rapid unfolding of information?

Texas has a number of gaps and barriers related to capacity and access to care, including for example:

- Limited and disparate distribution of genetic services providers,
- High number of federally designated health professional shortage areas,
- Large physical expanse and long distances between providers,
- Lack of public transportation in non-metropolitan areas,
- Growing ethnic diversity,
- Steady increase in births to women over the age of 35,
- Socioeconomic disparities,
- Limited insurance reimbursement rates, and
- High rate of uninsured individuals and families.

Texas is further challenged by the need to:

- Improve data collection, integration and reporting capabilities related to utilization of services, access to care, prevalence of genetic disorders, and efficacy of services.
- Enhance public understanding of genetics and its impact on overall health.
- Train primary care providers and other non-geneticist physician specialists to, at minimum, recognize indications for a genetic referral, know where to refer

patients needing genetic services, and work together with genetics professionals to coordinate and provide comprehensive care to individuals and families.

- Provide ongoing training for genetics specialists in order to integrate technology into clinical practice.

Recognizing the many challenges and opportunities ahead for Texas, the IACGS offers the following recommendations for consideration.

1. Increase access to genetic services in rural and underserved areas of the state.
    a. Allocate funds to implement telemedicine technology in outlying areas of the state. Resources needed include appropriate facilities, equipment, and technical assistance in the identification of professional partners and contract negotiation.
    a. Increase utilization of genetic counselors and advance practice nurses in genetic service delivery by allowing direct billing for their services.
    b. Strengthen primary care provider capacity through education and training to provide primary level genetic services.
    c. Provide financial assistance to students interested in becoming a physician geneticist or genetic counselor.

2. Allocate resources to develop and implement comprehensive genetics education and training at all levels, i.e., primary care and other non-geneticist physician specialists, nurses, medical and nursing students, physician assistants, physician geneticists, genetic counselors, and the general public.
    a. Identify, modify, as appropriate, and deliver existing education models.
    b. Collaborate with genetic service providers and professional education groups to develop web-based training opportunities for geneticist and non-geneticist health care providers, awarding appropriate continuing education credits per discipline. Information updates would be made to assure awareness of emerging issues and appropriate utilization of new genetic technologies.
    c. Utilize telemedicine as an education medium.
    d. Work with schools of public health, medicine, and nursing to promote the integration of genetics into professional study and practice.
    e. Work with lay and professional groups to modify existing or develop new language-appropriate education materials for consumers. Focus group test products and revise accordingly before release.

3. Promote access to family-centered, culturally, and linguistically appropriate genetic services.

a. Develop and employ recruitment strategies to attract diversity in genetics training programs.
b. Provide cultural competency information and training opportunities to genetic service providers.

4. Allocate funds to the IACGS to improve data collection and reporting capabilities about incidence of genetic disorders, and utilization and efficacy of genetic services for health resource planning and improvement.

5. Increase safety net program funding to address contract ceiling issues and to assist with plans for comprehensive education and training.

6. Increase efforts to assure adequate and timely reimbursement for genetic services.

INTRODUCTION

Genetic disorders, traditionally associated with problems of pregnancy and birth, are now more commonly recognized as occurring throughout the lifespan.

- Up to 70% of first trimester miscarriages are caused by chromosomal abnormalities.[1]
- Birth defects (congenital malformations) are the leading cause of infant death in the United States.[2] In Texas, 13,000 babies are born each year with one or more birth defects,[3] accounting for 25% of infant deaths.[4] Roughly one-third of birth defects are associated with a genetic cause.
- Almost 25% of hospital admissions for one-to-17 year-old Texas children in 2004 were due to a genetic disorder.[5]
- Nine of the 10 leading causes of death for all ages in Texas and nationally have genetic associations.[6]

What are genetic disorders?

Genetic disorders are those conditions or diseases associated with abnormalities or changes in genetic material (DNA). The disorders may be due to a missing or extra chromosome, an error in a single gene, or more commonly, the result of a complex interaction between multiple genes and environmental factors. Clinical presentation is variable, ranging from barely discernible to severely debilitating, but all involve abnormalities of structure and/or function. Genetic disorders typically are grouped into three categories: single-gene disorders, chromosomal abnormalities, and multifactorial inherited disorders. A small number of these conditions result from mutations in mitochrondrial DNA.

Single-gene, or Mendelian, disorders result from a mutation in the DNA sequence of a single gene. Single-gene disorders are very rare, but there are thousands of these

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[1] March of Dimes, *Chromosomal Abnormalities*, accessed from www.marchofdimes.com/professionals/681_1209.asp on August 1, 2006.

[2] Centers for Disease Control, National Center for Health Statistics (CDC/NCHS), *Number of infant deaths, %age of total infant deaths, and infant mortality rates for 2003: United States*, accessed from www.cdc.gov/nchs/data/hestat/finaldeaths03_tables.pdf on July 28, 2006.

[3] Department of State Health Services (DSHS), Birth Defects Epidemiology and Surveillance Branch, *About Birth Defects in Texas*, accessed from www.dshs.state.tx.us/birthdefects/about.shtm on August 4, 2006.

[4] DSHS, *Vital Statistics 2004 Annual Report, Table 32, March 27, 2006 and Table 31, January 12, 2006*, accessed from www.dshs.state.tx.us/CHS/VSTAT/vs04/t32.shtm and www.dshs.state.tx.us/CHS/VSTAT/vs04/t31.shtm on July 28, 2006.

[5] DSHS, Center for Health Statistics, *Hospital Inpatient Discharge Data, 2004.*

[6] CDC/NCHS, National Vital Statistics System, *Deaths, % of total deaths, and death rates for the 15 leading causes of death: United States and each State, 2003*, accessed from www.cdc.gov/nchs/data/dvs/lcwk9_2003.pdf on July 28, 2006.

disorders, making their cumulative effect more significant. Sickle cell disease, cystic fibrosis, phenylketonuria (PKU), Marfan syndrome, and Huntington's disease are examples of single-gene disorders. Symptoms range from mild to severe, may be present at birth or later in life, or may skip a generation, depending upon the pattern of inheritance. More severe effects include physical abnormalities, organ damage, mental retardation, and premature death.

Chromosomal abnormalities are due to an error in chromosomal number or structure. It is possible to inherit some types of chromosomal disorders, and it is possible for chromosomal abnormalities to occur in successive pregnancies, but most of these disorders are not passed down from generation to generation. The errors occur either during the formation of the egg or sperm, at conception, or during embryonic development. Effects can be profound, including miscarriage, stillbirth, and if the newborn survives, severe physical deformity, damage to multiple organs, mental retardation, and premature death, often within the first year of life. Other cases will be much less pronounced. Trisomy (extra chromosome) and translocation (rearrangement of chromosomes) disorders are among this category of genetic disease.

Multifactorial inherited disorders involve an interaction of one or multiple faulty genes with lifestyle and other environmental factors. In this case, we inherit a genetic susceptibility or predisposition to certain diseases. This "risk factor", when combined with environmental factors, such as maternal conditions during pregnancy, exposure to chemicals or infection, and behavior, will cause disease or maintain health. Multifactorial inherited disorders are the most commonly occurring category of genetic disorders and include many adolescent and adult-onset chronic diseases, such as some types of heart disease and cancer.

Historically, pregnant women, children, and infants have been the largest consumers of genetic services. While this still holds true, we are learning that an increasing number of commonly occurring diseases have a genetic causal component. Many of these do not appear until later in life. This has implications for the availability of genetic services throughout the lifecycle, and the integration of genetic services across medical specialties.

What are genetic services?

Genetic care involves the integration of clinical, laboratory, counseling, and follow-up services for individuals and families who have or are at risk for a disorder with a significant genetic component. The objectives of genetic care include identifying and mitigating problems, educating families to make informed decisions, and providing anticipatory care. Components include clinical evaluation and diagnosis, laboratory

testing to confirm diagnosis, genetic counseling, management and treatment of disorders, support and follow-up for individuals and families, accessible information for families, other health professionals and patient support groups, and referral to other medical specialists, social services, special education and support groups.

What are the benefits of genetic services?

- **Prevention or reduction** of adverse pregnancy outcomes through education and counseling. For example, women who are pregnant or considering pregnancy should be counseled to take adequate folic acid daily to protect babies from neural tube defects and to reduce the incidence of babies born with low birth weight. Similarly, women who could become pregnant should be advised to refrain from smoking and taking drugs, avoid exposure to infectious agents and environmental toxins, and remain under a physician's care for management of chronic illness.

- **Early diagnosis and treatment** of genetic conditions. A number of genetic disorders can be diagnosed before birth through the use of prenatal tests, and some may be treated. For example, biotinidase deficiency may be diagnosed through amniocentesis and treated with biotin vitamin supplementation, resulting in the birth of a healthy baby. Without treatment, this disorder can lead to seizures, developmental delay, and hearing loss. Texas currently screens newborns for five disorders, all of which may be treated through a combination of diet, medical formula, medications, and continued monitoring of the condition through blood, urine and other tests. In many cases, early detection and treatment will prevent long-term disability and premature death.

- **Risk assessment and predictive testing** for late onset disease. A number of disorders or diseases do not show recognizable signs until well into adolescence or adulthood. For example, symptoms of Huntington's disease (HD) often do not appear until an individual is in his/her thirties or older. However, we know that HD is a familial disease passed from parent to child through a gene mutation. The child of one parent with HD has a 50/50 chance of inheriting the defective gene. Presymptomatic testing is available for individuals who are at risk for carrying the HD gene.

- **Anticipatory care**. Individuals receiving care for a genetic condition may avert complications from secondary illnesses associated with the condition, through anticipatory care. For example, individuals with Williams syndrome will most likely have some type of heart or blood vessel problem. Typically, there is

narrowing in the aorta or in the pulmonary arteries. There is a broad range in the degree of narrowing, ranging from trivial to severe (requiring surgical correction of the defect). Since there is an increased risk for development of blood vessel narrowing or high blood pressure over time, periodic monitoring of cardiac status is necessary.

Another example of anticipatory care is the daughter who is concerned with developing breast cancer because her mother and maternal grandmother both had mastectomies due to breast cancer. The grandmother passed away, but the mother was tested and found to have a mutation of the gene BRCA2. The daughter tests positive for the same gene mutation, thereby increasing her risk of developing breast cancer. Knowing she has an increased risk, she schedules mammograms and clinical breast examinations every six months instead of yearly.

Anticipatory care is also seen in the child with Down syndrome whose parents enroll him in Early Childhood Intervention (ECI) because Down syndrome is associated with developmental delay.

- **Informed decision-making** goes hand-in-hand with anticipatory care in that the individual now has an accurate diagnosis, has become educated on the particular disease and its possible effects, has been counseled on available options and resources, and can make decisions within this framework.

Who provides genetic services?

Providers of genetic services include clinical physician geneticists, genetic counselors, and genetic laboratories. In addition, various aspects of genetic care are made available by obstetrician-gynecologists, family practice physicians, and other physician specialists who employ genetic screening tools and tests to assess patient risk for genetic complications.

Primary care physicians, other physician specialists, social workers, case managers, advanced practice nurses, and health educators also play a key role in providing education on prevention or risk factor reduction of genetic complications, as well as identifying and referring patients to a genetic specialist when indicated, and providing continuity of care. Since patient care and management of disorders is largely under the purview of primary care physicians, it is critical that they know when to refer their patients for genetic services. Table 1 lists some of the indicators for referral at the preconception/prenatal, pediatric, and adult stages of life.

TABLE 1. INDICATIONS FOR A GENETIC REFERRAL BY STAGE OF LIFE	
Preconception and Prenatal	▪ Maternal age ≥ 35 – associated with higher incidence of preterm and low birth weight babies; maternal complications, such as gestational diabetes and hypertension; problems associated with chromosomal disorders ▪ Maternal age ≤ 18 – associated with increased incidence of preterm and low birth weight babies ▪ Family history of genetic disorder ▪ Previous child with birth defect, chromosomal abnormality or other genetic disorder ▪ Birth defect or sign of genetic condition identified on ultrasound ▪ Abnormal serum screen for neural tube defects and chromosomal abnormalities ▪ History of miscarriage or stillbirth ▪ Exposure to infection, drugs, chemicals, toxins during pregnancy ▪ Carrier testing for specific ethnic groups who have a higher incidence of genetic disorders, e.g., sickle cell disease, Tay-Sachs disease
Pediatric	Newborns, infants, children with ▪ Birth defects or multiple congenital anomalies ▪ Known or suspected genetic disorders, such as Down syndrome, cystic fibrosis, muscular dystrophy ▪ Abnormal growth patterns, i.e., excessive growth, very short stature ▪ Abnormal body and limb proportions ▪ Ambiguous genitalia ▪ Abnormal or unusual facial features ▪ Developmental delays ▪ Mental retardation ▪ Metabolic disorders
Adult	▪ Abnormal growth patterns, i.e., excessively tall or short ▪ Adult onset genetic disorders, such as Huntington disease, Spinal Muscular Atrophy, Myotonic dystrophy ▪ Positive history of familial disorders, e.g., breast, ovarian and colon cancers, Duchenne's muscular dystrophy, Thalassemia, Tay-Sachs, sickle cell disease

In addition to family centered care, genetic services include population-based interventions, such as newborn screening and targeted health promotion campaigns, public health initiatives, such as surveillance and maintenance of disorder registries, education and training for health professionals, ongoing genetic research, and development of guidelines and standards for the provision of genetic services.

GENETIC SERVICES IN TEXAS:
AN INVENTORY OF RESOURCES

CLINICAL GENETIC SERVICE PROVIDERS

Clinical genetic services are medical services provided to individuals, families, and populations who have or are at risk for disorders with genetic implications. Services include testing, counseling, education, treatment, and where appropriate, referral for other services. Clinical genetic services are provided to Texans by both public and private entities, including university medical schools, hospitals, clinics, and private practices. Direct patient care is provided by a total of 48 American Board of Medical Genetics certified physicians and 69 Board certified counselors in Texas. Table 2 shows their distribution by Health Service Region (HSR) in relation to the region's population.

TABLE 2. GEOGRAPHIC DISTRIBUTION OF CLINICAL GENETICISTS IN TEXAS

Health Service Region	Texas Population[7]		Board Certified Physician Geneticists Providing Patient Care[8]		Board Certified Genetic Counselors[9]	
	Number	Percent	Number	Percent	Number	Percent
1	816,894	3.6%	0	0	2	2.9%
2/3	6,777,356	29.5%	7	14.6%	20	29%
4/5	1,833,829	8%	1	2.1%	0	0
6	5,416,840	23.5%	28	58.3%	33	48.5%
7	2,565,608	11.2%	2	4.2%	6	8.8%
8	2,321,016	10%	7	14.6%	5	7.4%
9/10	1,305,989	5.7%	0	0	0	0
11	1,965,023	8.5%	3	6.2%	2	2.9%
State Total	23,002,555	100%	48	100%	68	100%

In addition to providing patient care, many of the 48 physicians listed above also conduct research and teach at Texas medical schools. Still others who do not currently see patients are involved in research, administration, and teaching. The 48 physician geneticists are physically located in 12 Texas counties, with more than half located in Harris County. Similarly, 44% of the 68 genetic counselors are located in the Houston

[7] DSHS, Center for Health Statistics, *Projected Texas Population by County, 2005,* accessed from www.dshs.state.tx.us/chs/popdat/ST2005.shtm on August 28, 2006.
[8] American Board of Medical Genetics, accessed from www.abmg.org/genetics/abmg/verifications.htm on August 28, 2006.
[9] American Board of Genetic Counselors, accessed from www.abmg.org/genetics/abmg/verifications.htm on August 28, 2006.

area. HSR 1, a 41 county region in the Texas Panhandle, and HSR 9/10, an area consisting of 36 counties and covering 61,428 square miles in West Texas, have no physician geneticists in residence. These populations are served by physician geneticists who periodically travel to Amarillo, Lubbock, Abilene, El Paso, and San Angelo, and by two genetic counselors, one in Lubbock and one in Odessa.

HSR 4/5 covers East Texas counties and has a population of more than 1.8 million. A physician geneticist started practicing in Tyler in July 2006. Until that time, families in East Texas needing genetic services traveled either to Galveston, Houston, Dallas or Louisiana.

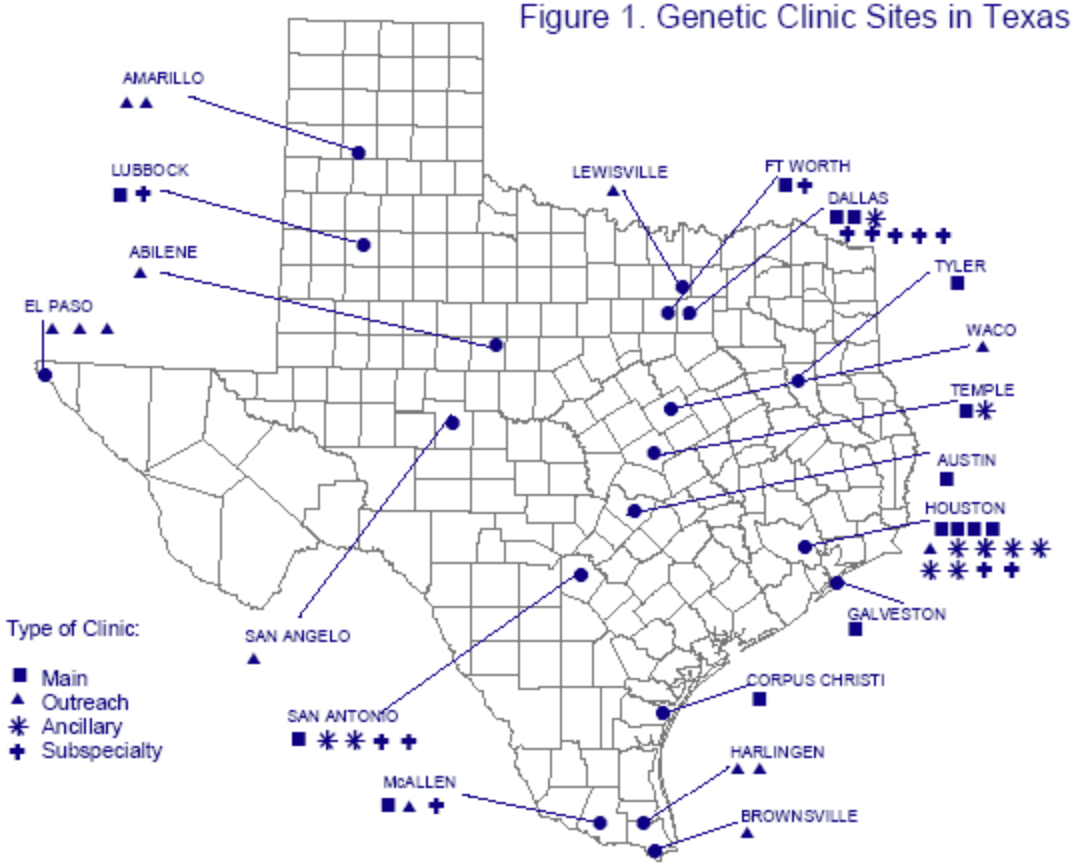
Poor distribution of health care providers is seen in other medical disciplines in Texas. For example, 27 counties had no primary care physician (PCP) and another 16 counties had only one PCP in 2005.[10] PCPs include family practice, general practice, internal medicine, general pediatrics, obstetrics and gynecology, and geriatrics. As of August 21, 2006, 116 Texas counties were designated as Primary Care Health Professional Shortage Areas (HPSA). Another 50 counties received partial designation as a HPSA.[11] The designation of a county as a HPSA indicates that the county has an inadequate number of specific health providers to serve the population in the county. Health Professional Shortage Areas are designated by the U.S. Department of Health and Human Services, Shortage Designation Branch.

The relative supply of PCPs and genetic specialists documents the disparity between various regions of the state in overall supply of health professionals and the need to address the issue of how to get more health professionals to practice in shortage areas to strengthen capacity and improve access to care.

[10] DSHS, Center for Health Statistics, Health Provider Resources Branch, *A Workforce Study of Medical, Nursing & Mental Health Professions in Texas*, May 9, 2006, accessed from www.dshs.state.tx.us/chs/hprc/workstu.pdf on July 28, 2006.

[11] DSHS, Center for Health Statistics, Health Professions Resource Center, *Primary Care HPSA Designations, Texas 2006,* accessed from www.dshs.state.tx.us/CHS/hprc/PChpsaWC.shtm and www.dshs.state.tx.us/CHS/hprc/PChpsaPT.shtm on August 29, 2006.

Figure 1. Genetic Clinic Sites in Texas



GENETICS CLINICS

Figure 1 illustrates the types and locations of clinics providing genetic services in Texas. There are a total of 49 clinics, two of which are classified as both a "main" clinic and a "subspecialty" clinic.

Main clinics generally have at least one full-time physician on staff and offer a full array of services, including medical evaluation and counseling for all ages and laboratory testing. An outreach clinic is an extension of a main clinic. It generally offers the same services in limited frequency. For example, the main clinic for Cook Children's Clinical Genetic Services is located in Fort Worth, where a full-time physician geneticist is housed. This physician travels to Cook Children's outreach clinics in Abilene, Amarillo, Lewisville, San Angelo and Waco, where she provides full services in limited frequency, e.g., one full-day clinic per month or quarter.

Ancillary clinics provide one or two genetic services on a regular basis. For example, Antoine Community Health Center and La Nueva Casa de Amigos, both located in Houston, provide genetic counseling only. Subspecialty clinics focus on a specific category of disorder or disease, e.g., cancer or metabolic disorders.

In Texas, there are 15 main clinic sites offering comprehensive services on a full-time basis. Only one main clinic is located in the vast area of the state west and north of San Antonio. There are 14 outreach clinics, providing full services on a limited frequency. There are 10 ancillary clinic sites offering limited services on a regular frequency. There are 12 subspecialty clinics, two of which are housed within a main clinic, offering services related to a specific disorder or set of disorders.

Consistent with health care provider distribution, clinics are sparsely located throughout most of the state, with clusters of clinics in key medical center areas: Houston, Dallas, and San Antonio. Families living in certain parts of the state, such as the Panhandle and West Texas, must travel large distances to access genetic services.

GENETICS LABORATORIES

Laboratories that provide genetic testing are either university, private, or state-operated, and concentrate on one or more of four areas of research: cytogenetic (chromosomal abnormalities), biochemical (inborn errors of metabolism), mitochrondrial (cellular energy), and/or DNA diagnostic (gene mutations).

<u>University Laboratories</u>

- Texas Tech University Health Sciences Center Cytogenetics Laboratory – Lubbock

- UT Southwestern Medical Center, McDermott Center for Human Growth and Development – Dallas
- UT Southwestern Medical Center, Veripath Laboratories -- Dallas
- University of North Texas Health Science Center – Ft. Worth
- Baylor College of Medicine, Kleberg Cytogenetics Laboratory – Houston
- Baylor College of Medicine, Biochemical Genetics Laboratory – Houston
- Baylor College of Medicine DNA Diagnostic Laboratory – Houston
- Baylor College of Medicine Mitochondrial DNA Laboratory -- Houston
- University of Texas MD Anderson Clinical Cytogenetics Laboratory – Houston
- University of Texas MD Anderson Molecular Diagnostic Laboratory -- Houston
- University of Texas Medical Branch Cytogenetics Laboratory – Galveston
- University of Texas Medical Branch Developmental Nutrition and Metabolism Laboratory -- Galveston
- Scott and White Cytogenetic Laboratory -- Temple
- University of Texas Health Science Center Clinical and Molecular Cytogenetics Laboratory – San Antonio

<u>Private Laboratories</u>

- Kimberly Courtwright and Joseph Summers Institute of Metabolic Disease at Baylor University Medical Center -- Dallas
- Center for Medical Genetics – Houston
- Dynagene – Houston
- Southwest Genetics PA – San Antonio

<u>State Laboratory</u>

- Department of State Health Services – Austin

POPULATION-BASED PROGRAMS

In addition to clinical services, Texas has population-based programs aimed at the early detection of disorders with a significant genetic association and referral to intervention services. Three programs are administered through the Department of State Health Services (DSHS), one program operates from the University of North Texas, and all are supported by state and federal funds.

- **Newborn Screening (NBS) Program**
  Operational since 1963, the NBS program was legislatively established to screen Texas newborns for five treatable genetic disorders. Effective January 2007, the program will screen for a total of 27 disorders, which if diagnosed and treated early in life, may prevent severe mental retardation, illness, and/or death.

All babies born in Texas are required to have two panels of blood screening tests. The DSHS Laboratory receives and analyzes more than 3,000 specimens daily. Abnormal results are immediately communicated to case management staff who provide notification and follow-up to the baby's family and physician. Table 3 shows the number of newborns screened at least once for five disorders, results, and follow-up for Fiscal Year (FY) 2005.

TABLE 3. NUMBER AND PERCENTAGE OF NEWBORNS SCREENED, CONFIRMED AND TREATED[12]

Total Births by Occurrence: 388,394* Reporting Year: FY05 (Sept 1, 2004 Thru Aug 31, 2005

Screening Tests	Receiving at least one Screen		Number of Presumptive Positive Screens	Number of Confirmed Cases	Babies Needing Treatment Who Received Treatment	
	Number	Percentage			Number	Percentage
Phenylketonuria (Classical)	374,682	96.46	215	2	2	100
Congenital Hypothyroidism (Primary)	374,682	96.46	7,500	240	240	100
Galactosemia (Classical)	374,682	96.46	841	1	1	100
Sickle Cell Disease	374,682	96.46	134	134	134	100
Congenital Adrenal Hyperplasia (Classical)	374,682	96.46	2,921	49	49	100

*FY05 births by occurrence is a preliminary number and is subject to change.

A total of 11,611 babies were identified with presumptive positive screens and referred for confirmatory testing. A total of 426 were confirmed as having a genetic disorder, all of whom received treatment.

[12] DSHS, *FY05 Title V Annual Report.*

- **Texas Early Hearing Detection and Intervention (TEHDI) Program**
  Following a successful pilot project, DSHS was legislatively mandated in 1999 to provide newborn hearing screening for Texas newborns prior to hospital or birth center discharge. Historically, children typically were not identified with hearing loss until they were 24 to 30 months of age. However, language development and cognitive outcomes are improved if treatment and intervention begins by six months of age.[13] [14]

  TEHDI provides oversight to 235 participating hospitals and birth centers that screen newborns for hearing loss and report data to the state. In calendar year 2005, 336,379 newborns were screened for hearing loss. That number represents 98.4% of live births at participating birth facilities. Just over three percent, or 10,988 babies were referred for follow-up screening and evaluation. About 1,000 will receive a confirmed diagnosis of moderate to profound hearing loss.[15]

  Moderate to profound bilateral hearing loss is estimated to occur in three of 1,000 births. This ratio increases to five to six infants per 1,000 births if mild and unilateral cases are included.[16] Approximately 50% of congenital or early-onset hearing loss is due to genetic causes. The other 50% is associated with environmental factors, such as maternal infection, prematurity, or exposure to certain drugs or chemicals. More than 400 different forms of hereditary hearing loss have been identified.[17] [18]

  Hearing loss may have delayed onset. In fact, hearing loss can appear at any age and with any degree of severity. Children who have genetic syndromes associated with hearing loss or other indicators for delayed onset or progressive hearing loss should receive annual hearing tests or be tested when symptoms occur in order to ensure prompt identification and treatment.[19] In addition to TEHDI, DSHS administers the **Vision and Hearing Screening Program**, a

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[13] Joint Committee on Infant Hearing. *Year 2000 Position Statement: Principles and Guidelines for Early Hearing Detection and Intervention Programs,* Pediatrics Vol. 106, No. 4, October 2000.
[14] Finitzo PhD, T., Albright MS, K., and O'Neal MFA, J. *The Newborn With Hearing Loss: Detection in the Nursery,* Pediatrics, Vol. 102, No. 6, December 1998.
[15] DSHS, Texas Early Hearing Detection and Intervention Program, *TEHDI Data Reporting System,* August 31, 2006.
[16] CDC, National Center on Birth Defects and Developmental Disabilities, *Early Hearing Detection and Intervention Program,* accessed from www.cdc.gov/ncbddd/ehdi/ehdi.htm on September 1, 2006.
[17] Burton, S.E., Pandya, A., and Arnos, K.S. *Genetics and hearing loss: An overview,* The ASHA Leader, January 17, 2006, 11(1), 1, 32, accessed from www.asha.org/about/publications/leader-online on September 1, 2006.
[18] American College of Medical Genetics. *Genetics Evaluation Guidelines for the Etiologic Diagnosis of Congenital Hearing Loss,* Genetics in Medicine, May/June 2002, Vol. 4, No. 3.
[19] Joint Committee on Infant Hearing. *Year 2000 Position Statement: Principles and Guidelines for Early Hearing Detection and Intervention Programs,* Pediatrics Vol. 106, No. 4, October 2000.

school-based detection and referral program for children with vision or hearing problems.

- **Texas Birth Defects Registry**
  The Texas Birth Defects Registry was established in 1994 to identify and describe the patterns of birth defects in Texas through active surveillance. The Registry monitors all births in Texas and identifies cases of birth defects. For the four-year period 1999 to 2003, the Registry reported a total of 65,911 infants and fetuses with any monitored birth defect, a prevalence of 360.7 per 10,000 live births.[20] Children identified through the Registry are referred to appropriate medical and community services. Additionally, researchers use Registry data to conduct epidemiological studies to find preventable causes of birth defects in Texas.

- **Texas Teratogen Information Service (TTIS), University of North Texas**
  Adverse pregnancy and birth outcomes may be prevented or reduced by avoiding or minimizing exposure to teratogens during the prenatal period. A teratogen is any chemical (herbicides, industrial solvents, lead, mercury), substance (cigarette smoke, drugs, alcohol), infectious agent (rubella, toxoplasma) or maternal condition (diabetes, maternal PKU) that may cause injury to the developing embryo or fetus during pregnancy. Effects are relative to the type of agent, dose, duration and timing of the exposure, but can include miscarriage, stillbirth, preterm delivery, low birth weight, birth defects, developmental delay, mental retardation, failure to thrive, and death.

  Well-known examples of teratogens include rubella and thalidomide. Infection with rubella during the first trimester of pregnancy is associated with a high risk of birth defects, including hearing loss, heart defects, mental retardation, and slow growth. Thalidomide was widely prescribed in the late 1950s for adverse symptoms of pregnancy, such as morning sickness. After thousands of babies were born worldwide with severe limb defects, thalidomide was banned from use. However, it is currently being used in the treatment of multiple myeloma and Hansen's disease.

  The Texas Teratogen Information Service (TTIS) at the University of North Texas is a statewide counseling and information program for individuals who have

[20] DSHS, Birth Defects Registry, *Report of Birth Defects Among 1999-20003 Deliveries,* accessed from www.dshs.state.tx.us/birthdefects/Data/reports.shtm on September 1, 2006.

questions or concerns about exposures to teratogens during pregnancy. TTIS also serves as a resource and distributes information to health care providers.

REHABILITATIVE AND INTERVENTION PROGRAMS

An important element of genetic services is patient referral to appropriate medical specialists and community based support programs as indicated. Texas offers a number of intervention and rehabilitative programs operated by its health and human service agencies.

Department of Assistive and Rehabilitative Services (DARS)

- **Early Childhood Intervention (ECI)** – state and federally funded through the Individuals with Disabilities Education Act (IDEA, PL 108-446), ECI provides an array of services for children birth to 3 years old with developmental delay, atypical development, or a medical diagnosis with a high likelihood of developmental delay. Families and professionals work together to plan appropriate services for each child. Services are provided in the home and in community settings such as child care facilities, play groups and Mothers' Day Out programs. ECI coordinates services that may include: occupational therapy, physical therapy, speech-language therapy, vision services, nutrition services, assistive technology services, and family counseling, among others. Families enrolled in Medicaid or CHIP, or with incomes below 200% of the Federal Poverty Income Level do not pay for services.[21] Others pay a cost share for services based on a sliding fee scale.

  In FY 2004, ECI provided comprehensive and follow along services to more than 49,000 children through its provider network at school districts, regional education service centers, community mental health and mental retardation centers, and private nonprofit organizations.[22]

- **Hard of Hearing Services** – available for persons of all ages who are hard of hearing, late-deafened, or oral deaf through contracted services with local providers. Services include information, referral, training, adaptive equipment demonstrations, and may include services of a hearing specialist.

[21] Department of Assistive and Rehabilitative Services (DARS), *ECI Services and Eligibility,* accessed from www.dars.state.tx.us/ecis/eligibility.shtml on September 7, 2006.
[22] DARS, *ECI Fact Sheet,* accessed from www.dars.state.tx.us/ecis/factsheet.shtml on September 7, 2006.

- **Services for the Blind and Visually Impaired** – provides vocational and independent living training to persons of all ages who are blind or visually impaired, and helps children receive the necessary training to be successful in school and beyond. The Transition Program, geared to 10 to 24 year-olds, includes: vocational training, orientation and mobility instruction, reader services, transportation, technological aids and devices, and employment assistance.

- **Comprehensive Rehabilitation Services** – helps persons with spinal cord and brain injuries receive therapies to increase independence.

- **Deafblind Services** – provides services to persons with deafblindness, including employment assistance, adaptive equipment, and establishing communication systems.

- **Vocational Rehabilitation Program** – assists people with physical and mental disabilities prepare for, find, and retain jobs. Work related services are individualized and may include counseling, training, medical treatment, assistive devices, and job placement assistance.

Department of State Health Services (DSHS)

- **Children With Special Health Care Needs (CSHCN) Services Program** – provides health benefits and case management to individuals younger than 21 who have a chronic physical or developmental condition, and to individuals of any age who have cystic fibrosis.[23] The program contracts with community-based organizations throughout the state to provide clinical and support services to children with extraordinary medical needs, disabilities, and chronic conditions, and their families. Program health benefits include: primary and preventive care, speech and hearing services, vision care, mental health services, ambulance, hospital care, medicines, special nutritional products, care by medical specialists, home health nursing, physical and occupational therapy, meals and lodging when needed to obtain medical care, and other support services.[24] Medicaid, CHIP, or other coverage must be used before CSHCN benefits.

[23] DSHS, *Children With Special Health Care Needs Program Health Benefits,* accessed from www.dshs.state.tx.us/cshcn/benefits.shtm on September 7, 2006,
[24] Ibid.

- **Program for Amplification for Children of Texas (PACT)** – serves Medicaid and CSHCN eligible Texas children from birth through age 20 with permanent hearing loss. Services include evaluation by an audiologist or otologist, hearing aid evaluation, earmolds and hearing aids, hearing aid follow-up visits, hearing aid repairs, earmold replacements, and hearing aid replacements when the current aids are no longer appropriate or are five years old.[25] PACT provides about 3500 hearing aids each year to Texas children.[26]

Department of Aging and Disability Services (DADS)

- **Services for Children** – DADS provides a number of in-home and community-based services so that children with disabilities may remain in their own homes or in other permanent community living arrangements. Services are designed to address the physical, mental, medical, and social needs of clients. Two programs, Community Living Assistance and Support Services and Medically Dependent Children, had waiting lists of 15,009 and 10,111, respectively, as of June 30, 2006.[27]

- **Services for Persons with Mental Retardation** – DADS provides services to individuals with mental retardation through 41 community mental health/mental retardation centers, waiver programs, intermediate care facilities, and state facilities. Medicaid home and community-based waiver programs provide services, such as adaptive aids, minor home modifications, nursing, counseling, respite, and supported employment, to individuals with mental retardation who either live with their family, in their own home, or in a residence with no more than four individuals who receive services. Intermediate care facilities provide residential and habilitative services, skills training, and 24-hour supervision. State facilities include 11 state schools and one state center, which provide 24-hour supervision and treatment. The Home and Community-based Services waiver program had a waiting list of 30,398 as of June 30, 2006.[28]

[25] DSHS, Audiology Services, *Program for Amplification for Children of Texas,* accessed from www.dshs.state.tx.us/audio/program.shtm on September 7, 2006.
[26] Ibid.
[27] Department of Aging and Disability Services, *Interest List Reduction Report Summary Fiscal Years 2006-2007*, June 30, 2006, accessed from www.dads.state.tx.us/services/interestlist/index.html#summary on September 7, 2006.
[28] Ibid.

FUNDING SOURCES

Genetic services in Texas are paid for by both public and private means. Three jointly funded state-federal programs, Medicaid, the Children's Health Insurance Program (CHIP), and the Title V Maternal and Child Health Block Grant are programs that primarily serve low-income families.

Medicaid

The Texas Medicaid program was established in 1967 and since 1993 has been administered by the Health and Human Services Commission (HHSC). Medicaid is an entitlement program, which means neither the federal nor state government can limit the number of eligible people who can enroll and receive services covered under the program. In FY 2004, 2.9 million Texans were enrolled in Medicaid at an estimated cost of $17 billion.[29] [30] Two-thirds of enrollees were under age 20, and 83% of enrollees lived at or below the federal poverty income level, earning less than $20,000 for a family of four.[31]

Medicaid covers three primary groups:

1. Families and children – based on income level and depending on age or pregnancy status. Families and children represent 62% of the total Medicaid caseload.[32]

2. Cash assistance recipients – based on receipt of Temporary Assistance to Needy Families (TANF) or Supplemental Security Income (SSI). This category accounts for 28% of the total caseload.[33]

3. Aged and disabled – based on income level, age, and physical or mental disability. Aged and disabled represents 10% of the Medicaid caseload.[34]

Texas Medicaid providers are reimbursed through either the traditional fee-for-service model or through one of two types of managed care plans: health maintenance

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[29] Health and Human Services Commission (HHSC), *Medicaid in Perspective, June 2004, 5-1,*accessed from www.hhsc.state.tx.us/medicaid/reports/PB5/PDF/Chapter05.pdf on September 9, 2006.
[30] HHSC, *Medicaid Monthly Enrollment History as of October 2005*, accessed from www.hhsc.state.tx.us/research/dssi/medicaid/McaidHist2.asp on September 11, 2006.
[31] Texas Medical Association (TMA), *Medicaid: 40 Years of Helping Texans Get Access to Health Care, July 2005,* accessed from www.texmed.org/printthispage.aspx?id=4168 on September 11, 2006.
[32] HHSC, *Medicaid in Perspective, June 2004, 4-4,*accessed from www.hhsc.state.tx.us/medicaid/reports/PB5/PDF/Chapter04.pdf on September 12, 2006.
[33] Ibid.
[34] Ibid.

organization (HMO) or primary care case management (PCCM). HMOs are licensed by the Texas Department of Insurance and deliver services under a risk-based arrangement. These organizations receive a monthly capitation payment for each person enrolled based on an average projection of medical expenses for the typical patient.[35] In the PCCM model, each Medicaid client is assigned a primary care provider (PCP). The PCP must authorize most other health care services, such as specialty physician care, before Medicaid will pay for them. Providers receive the fee-for-service reimbursement rate.[36]

The federal government defines certain mandatory services each state must provide, e.g., inpatient and outpatient hospital services, physician services, lab and X-ray, immunizations and other well child services, and an array of optional services from which the state may choose to provide. Texas provides a number of optional services, including prescription drug coverage, hearing services, physical and occupational therapy, and genetic services, among others.[37]

DSHS oversees the provision of genetic services reimbursable by Medicaid. Providers enrolled as Medicaid approved genetic service providers must meet certain requirements. For example, the provider's medical director must be a clinical geneticist (MD or DO) who is licensed by the Texas Medical Board and certified by the American Board of Medical Geneticists (ABMG), and the laboratory used for confirmatory testing must be certified under the Clinical Laboratory Improvement Amendments (CLIA), Centers for Medicare and Medicaid Services.[38] Genetic providers are reimbursed according to an established allowable maximum fee schedule to evaluate, diagnose, counsel, and follow-up with clients with known or suspected genetic disorders. Services may include genetic history and physical examination, psychosocial genetic assessment, laboratory services, radiological services, diagnostic procedures, and counseling.

There are four key limitations with the current system: the reimbursement rate, the number of procedure (CPT) codes that are reimbursable, the frequency in which services may be reimbursed, and limited eligibility for particular procedures. Maximum Medicaid reimbursement rates are typically 50-70% of established Medicare

[35] Texas Comptroller of Public Accounts, *Texas Health Care Claims Study, March 2005,* accessed from www.window.state.tx.us/specialrpt/hcc2005/sect1a.htm on September 12, 2006.
[36] Texas Comptroller of Public Accounts, *Texas Health Care Claims Study, March 2005,* accessed from www.window.state.tx.us/specialrpt/hcc2005/sect1a.htm on September 12, 2006.
[37] Ibid.
[38] Texas Medicaid & Healthcare Partnership, *2006 Texas Medicaid Provider Manual, 23:2-14.*

rates.[39] Additionally, there are limitations on the number of times a procedure may be billed per client, e.g., "one per lifetime per provider" or "two per provider per lifetime of client per specimen." Regarding limited eligibility for procedures, Medicaid will cover certain tests for mothers and children, but may not cover these tests for fathers, as illustrated in the following case study.

> *Case Study. A pregnant woman has an abnormal triple screen. She is referred to an OB-GYN or geneticist for diagnostic testing. The tests may include amniocentesis with a chromosome study or a high-resolution ultrasound. The Medicaid provider performs amniocentesis, which reveals the baby has a chromosome rearrangement. This may have been inherited from a normal parent, i.e., showing no chromosomal abnormalities, and may not be indicative of a problem. In order to determine if there is a problem, both parents should receive chromosome testing. Medicaid will pay for the mom to be tested, but will not cover the dad.*

In the case study above, the "dad" is seasonally employed and does not have other insurance coverage. If he is to have the test, he will have to pay for it himself.

Children's Health Insurance Program (CHIP)

CHIP was established in 1999 and is administered by the HHSC. It is a health insurance program available to children whose families earn too much money to qualify for Medicaid, but cannot afford private insurance. Most families pay premiums and co-payments based on income and assets and the number of people in the family. Enrollment fees do not exceed $50 for each six-month term of eligibility and co-payments for doctor visits and prescription medicines range from $3 to $10.[40]

To qualify for CHIP, a child must be under age 19, a Texas resident, and a U.S. citizen or legal permanent resident. Services are offered through private health maintenance organizations (HMO) at contracted rates. CHIP provides coverage for a full range of health services including regular checkups, immunizations, prescription drugs, lab tests, X-rays, hospital care, and specialty care upon referral. Because of the limited number of clinical geneticists in Texas or because long distance travel is a barrier, a

[39] Zuckerman, S., McFeeters, J., Cunningham, P., and Nichols, L. *Changes in Medicaid Physician Fees, 1998-2003: Implications for Physician Participation,* Health Affairs, Web Exclusive, June 23, 2004.

[40] HHSC, *A Consumer Guide for Better Health, 2005: Your Guide to Understanding the Health Care System in Texas,* accessed from www.hhsc.state.tx.us/chip/reports/ConsumerGuideEnglish.pdf on September 13, 2006.

patient may need to see an out-of-network provider. In these cases, reimbursement for claims is at the discretion of the HMO. Some genetic service providers have reported difficulty in getting payment, either having the claim rejected or only receiving partial payment.

The new CHIP Perinatal program is due to roll out in January 2007. CHIP Perinatal is designed to provide prenatal care to the unborn children of women who do not qualify for Medicaid, including legal permanent immigrants and undocumented immigrants.[41] Benefits include up to 20 prenatal visits, limited laboratory testing, prescription drugs, education and counseling, and two postpartum visits for the mother.

Title V Genetic Services

Title V of the Social Security Act pledges federal support to states in the form of a Maternal and Child Health (MCH) Block Grant with state matching funds. The state MCH block grants are intended to provide and assure mothers and children access to quality maternal and child health services, and in particular, to provide prenatal, delivery and postpartum care for low income, at risk pregnant women and preventive and primary care services for low income children.

The DSHS Genetic Services program contracts with university medical schools, physician practices affiliated with not-for-profit hospitals, and private practice physicians to provide clinical genetic services to Title V eligible clients. These services are provided to Texas residents with family incomes at or less than 185% of the most current Federal Poverty Income Level, who are not eligible for other programs providing the same services. Contractors provide specified allowable services at Medicaid established rates on a fee-for-service basis. Once contract ceilings are reached, no further funds are allocated. This poses some problems.

A number of contractors reach contract ceiling limits at two-thirds of the contract year, and either do not continue to provide services to Title V eligible clients or provide charity services. Additionally, contractors must decide how to best use their limited Title V funds. In the case study above of the woman who presented with a positive triple screen, further testing by amniocentesis with a chromosome study or a high-resolution ultrasound was indicated. The Title V contractor, who is fast approaching his contract ceiling must decide whether to spend part of his allocation on one of these tests or instead, for example, provide medical genetic counseling to a previously unserved family. These problems are further compounded by the limited availability of

[41] TMA, *2005 Legislative Compendium: Health Care Funding,* accessed from www.texmed.org/printthispage.aspx?id=4233 on July 5, 2006.

Title V funds for genetic services, just short of $1.2 million per year. A portion of that stipend is allocated to a population-based education program. In FY 1997, dollars allocated to Title V genetic services were $1,834,134.

In FY 2005, some 4954 unduplicated clients received clinical genetic services through Title V, with another 615 people reached through the population-based Texas Teratogen Information Service. In FY 2006, there are a total of eight contracted providers of clinical genetic services and one population-based contracted program. The clinical service providers are located in: Austin, Corpus Christi, Fort Worth, Galveston, Houston, Lubbock, and McAllen. In order to partially fill in the large gap left in West Texas, DSHS provides genetic services at its El Paso clinic for three days each month.

Prior to 1996, DSHS, then the Texas Department of Health, operated genetics clinics at 22 locations throughout the state. The program was headquartered in Denton and from that location, geneticists traveled to the clinic locations to provide services to low income families. Funding was provided from a general revenue appropriation and generation of third-party user fees.[42] In early 1996, the genetics clinics were closed, with the exception of the one in El Paso. Instead, the provision of genetic services to low income families would be accomplished through a competitive grant application process, with Title V funds awarded to private and public genetics providers.

Private Health Care Coverage

Roughly 58.5% of non-elderly Texans received health care coverage through private health insurance in 2005, most often through their employers.[43] Ironically, while most individuals aged 18-65 who have health insurance access it through their place of employment, almost two-thirds of those who are uninsured are employed.[44] These uninsured adults either work in jobs that do not provide health insurance or are unable to afford the coverage offered. Seven (7) percent of privately insured Texans purchase their own health insurance plans.

Another idiosyncrasy related to health insurance is that private insurance tends to cover healthy individuals. Many of the sickest and most expensive patients are those who are unable to work, have met their lifetime benefit, have pre-existing conditions for which

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[42] Texas State Genetics Plan Advisory Council Executive Committee, *The Development of the Texas State Genetics Plan and a Plan for Integrated Data Infrastructure for Genetic Services, 2002.*

[43] U.S. Census Bureau, Historical Health Insurance Tables, *Health Insurance Coverage Status and Type of Coverage by State: 1987-2005,* accessed from www.census.gov/hhes/www/hlthins/historic/hihistt4.html on September 8, 2006.

[44] HHSC, *Medicaid in Perspective, June 2004,2-5,* accessed from www.hhsc.state.tx.us/medicaid/reports/PB5/PDF/Chapter02.pdf on September 12, 2006.

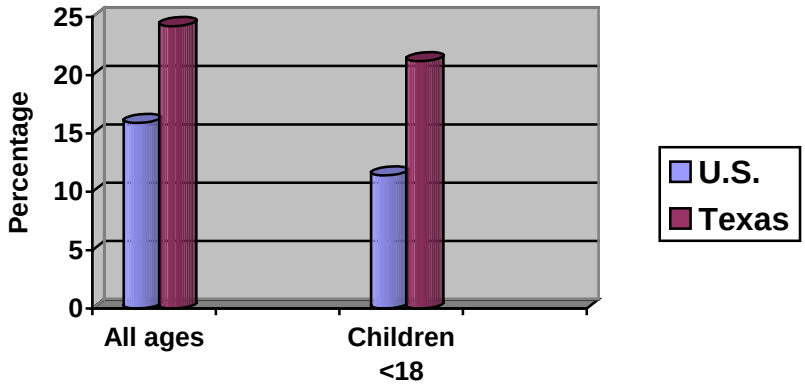
treatment is excluded for up to 12 months, or cannot afford insurance premiums.[45] Costs for their care are often transferred to the public sector and supported by federal, state, or local governments.

No Health Care Coverage

According to the U.S. Census Bureau, almost one in six Americans is uninsured; in Texas, the ratio is one in four. While there is variability among Texas counties, every major Texas city has an uninsured rate that is higher than the national average.[46] In fact, Texas leads all states in the percentage of people who are uninsured.[47] Texas' share of uninsured children is also higher than the U.S. average. Between 2002 and 2003, 21.2% of Texas children under the age of 18, were uninsured, compared to 11% nationally.[48]

Figure 2 compares uninsured rates in the United States and Texas.

Figure 2. Percentage of Population Without Health Insurance, U.S. and Texas, 2005



Source: U.S. Census Bureau, Historical Health Insurance Tables, 1987-2005.

The uninsured include diverse groups of people: those who cannot afford private health insurance; who work in small businesses that do not offer insurance; who are eligible for government-sponsored programs, such as Medicaid, CHIP or Title V, but are not enrolled; and even those who can afford insurance but choose not to purchase it.

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[45] Ibid, *2-6.*

[46] Texas Comptroller of Public Accounts, *The Uninsured: A Hidden Burden on Texas Employers and Communities, April 2005,* accessed from www.window.state.tx.us/specialrpt/uninsured05/ on September 11, 2006.

[47] U.S. Census Bureau, *Income, Poverty, and Health Insurance Coverage in the United States: 2005,* accessed from www.census.gov/prod/2006pubs/p60-231.pdf on August 24, 2006.

[48] State Health Access Data Assistance Center and the Urban Institute, *Going Without: America's Uninsured Children, August 2005,* accessed from http://www.rwjf.org/files/newsroom/ckfresearchreportfinal.pdf on October 5, 2006.

Health insurance coverage is an important determinant of access to health care. Without it, people are less likely to seek preventive care, have a medical home or regular place to go to for medical care, and more likely to use emergency rooms for non-urgent care. Other factors affecting access to health care include: adequate supply of health care professionals, geographic location of providers, cultural competency of providers, and affordability of health care services, with or without health insurance.

GENETIC SERVICES IN TEXAS:
CHALLENGES AND OPPORTUNITIES

Limited workforce capacity, access to care, and funding are inherent problems in most state health care systems. These problems are exacerbated in Texas by its particular geography and demography.

TEXAS GEOGRAPHY AND DEMOGRAPHY

The physical expanse of Texas in itself poses a major challenge to accessing health care. Second in size only to Alaska, Texas occupies an area as large as all of New England, New York, Pennsylvania, Ohio, and North Carolina. The greatest north to south distance in the state is 800 miles, and the largest east to west distance is 773 miles.[49] Individuals and families needing genetic services often must travel great distances to see a genetic specialist. Texas' limited supply of genetic physicians and counselors are highly concentrated in HSR 6, with no genetic physicians residing in HSR 1 or 9/10, and one only recently established in HSR 4/5. Similarly, genetic clinic sites are sparsely located throughout the state, with the heaviest concentrations in Houston, Dallas, and San Antonio.

While some geneticists provide periodic clinics in outlying areas, these occur in limited frequency. Patients may wait a month or more for the next clinic to be held near where they live, or travel hundreds of miles to see a genetic physician sooner, often missing work, as well as incurring costs of travel and lodging. This is further complicated by limited public transportation outside of metropolitan areas. The majority of Texas counties are rural or non-metropolitan, and include a large number of counties bordering Mexico.

Texas' population is second in size only to California, and similarly diverse in its ethnic distribution. In 2004, minority populations, collectively, surpassed the size of the non-Hispanic white population in Texas.[50] In 2026, the Hispanic population on its own is projected to become the majority population in Texas. This has implications for the cultural competency of genetic specialists and health care providers in general, already identified as a concern in previous Resource Allocation Plans.

Texas leads the nation in the percentage of people who do not have any type of health insurance coverage. This is further complicated by its high percentage of people living

[49] 2006-2007 Texas Alamanac, accessed from www.texasalmanac.com/envrionment/ on August 30, 2006.
[50] DSHS, Office for the Elimination of Health Disparities, *Health Disparities Task Force Annual Report, 2004.*

in poverty. In 2004, 16.7% of Texans were living at or below the Federal Poverty Income Level.[51]

Another significant trend nationally and in Texas is the increasing number of births to women who are 35 years of age or older. As table 4 illustrates, Texas has seen a steady increase in births to women in this age group.[52]

TABLE 4. BIRTHS TO TEXAS WOMEN ≥ 35, 2000-2003					
Year	2000	2001	2002	2003	2000-2003
Births	37,723	37,971	39,082	40,339	155,115

	2000-2001	2001-2002	2002-2003	2000-2003
% Increase in ≥ 35 Births	0.7%	2.9%	3.2%	**6.93%**

The March of Dimes explains that women over the age of 35 have a higher likelihood of pregnancy complications including: gestational diabetes, placental problems, miscarriage, premature delivery, stillbirth, or having a child with certain birth defects.[53] Preconception counseling and prenatal care are indicated for this age group. The American College of Obstetricians and Gynecologists recommends that women who will be 35 or older at the time of delivery be offered prenatal testing to rule out possible chromosomal abnormalities.

CHANGES OVER THE YEARS

In addition to geographic and demographic challenges, Texas has experienced operational changes over the years, which have impacted its genetic service capacity and access to care. In 1996, the former Texas Department of Health's Genetic Screening and Counseling Service, which was based in Denton and oversaw the provision of genetic services at 22 clinics, was discontinued. Instead, through a competitive bid process, contracts were awarded to physician geneticists in public or private practice to provide genetic services to Title V eligible clients. This reduced the number of genetics clinics throughout the state, diminishing capacity and access.

In FY 1997, the former Texas Department of Health (now DSHS) awarded over $1.8 million in Title V funds to contractors. This included more than $1.3 million in direct patient services and just under $500,000 in education and population-based services. This amount has been reduced over the years, with current contract awards totaling

[51]U.S. Census Bureau, Current Population Survey, *2004-2006 Annual Social and Economic Supplements.*
[52]DSHS, Research and Public Health Administration, 2006.
[53] March of Dimes, *Pregnancy After 35,* accessed from www.marchofdimes.com/printableArticles/14332_1155.asp on September 5, 2006.

$1.2 million. This funds eight fee-for-service providers and only one population-based program, impacting capacity, access, and the availability of educational information and outreach.

In 2001, TexGene was discontinued. Operational from 1994 to 2001, TexGene was a collaborative group of genetic service providers, who with funds from the federal Health Resources and Services Administration (HRSA), surveyed and collected data from Texas providers. The data collected through these surveys included such information as number of patients served and patient demographics, reason for referral and services provided, method of payment for services, and information about the provider. The analysis of this data by the DSHS Research and Public Health Assessment Office (RHPA) provided important service utilization information, presented in Resource Allocation Plans from 1996 through 2006, with the later reports using projections. Funding for this project also helped fulfill the Interagency Council for Genetic Services' (IACG) objectives: to survey current resources for human genetic services in the state, evaluate current and future needs for services, assist in coordinating statewide human genetic services, and monitor the provision of human genetic services. Without the funding of approximately $20,000 biennially, the IACGS is unable to continue these functions.

NBS EXPANSION

House Bill 790, 2005 (79(R)), directed DSHS to expand its NBS program. Texas currently screens for five heritable disorders. The expanded program, due to roll out in January 2007, will screen for 27 disorders, as recommended by the American College of Medical Geneticists. In FY 2005, 11,611 newborns were identified with presumptive positive screens and referred for confirmatory testing. A total of 426 were confirmed as having a genetic disorder and referred for treatment.[54] Increasing the panel from five to 27 disorders is anticipated to increase the number of presumptive positive screens to 15,000 or by more than 25%.

As a result of the NBS expansion, Texas newborns will be screened for many additional disorders, all of which are treatable. These disorders, if diagnosed and treated early in life, may prevent severe mental retardation, illness, and/or death. The expansion also impacts providers. An increase in presumptive positive screens should result in an increase in confirmatory testing. An increase in confirmed cases of genetic disorders will likely result in an increase in the utilization of genetic services.

[54] DSHS, *FY05 Title V Annual Report.*

The NBS expansion has resulted in the need for additional DSHS laboratory and follow-up/case management staff, as well as educational materials and training for providers and the public.

HUMAN GENOME PROJECT (HGP)

The Human Genome Project, completed in 2003, was a 13-year international effort coordinated by the U.S. Department of Energy and the National Institutes of Health, whose goal was to determine the complete structure of the human genome (the entire set of genes of an individual) and understand its function. Information gleaned from this project would be used for continued biomedical research aimed at better understanding of disease processes, and their prevention, detection, diagnosis, management and treatment. Some applications of genome research include earlier detection of genetic predispositions to disease, improved diagnosis of disease, and tailored treatments, including pharmacogenomics and gene therapy. Research has led to the availability of numerous genetic testing capabilities and increased demand by consumers for genetic information and advice. This has challenged an already taxed genetics workforce and is changing traditional boundaries between medical disciplines.

As genetic medicine becomes more pervasive, primary care providers and other non-geneticist specialists may become increasingly involved in:

- Identifying individuals who may benefit from genetic services, including those with a genetic disorder and those at increased risk for having or transmitting a genetic disorder,
- Recognizing physical and historical features of genetic disorders,
- Ordering and interpreting genetic predictive tests,
- Providing basic genetic information and counseling to facilitate informed decision-making,
- Knowing the full range of genetic specialists available in one's area and when referral and collaboration are indicated, and
- Coordinating care for individuals with complex genetic service needs.[55]

Clinicians and other health professionals will need to integrate genetics knowledge, skills, and attitudes into routine health care, in an effort to provide effective and comprehensive services to individuals and families. Recognizing this need, the National Coalition for Health Professional Education in Genetics (NCHPEG) developed a set of core competencies in genetics to provide a framework from which instructional

[55] Emery, J and Hayflick, S, *The challenge of integrating genetic medicine into primary care,* BMJ 2001; 322:1027-1030, April 28, 2001.

materials and educational programs have been and continue to be developed.[56] The NCHPEG says that at a minimum, each health-care professional should be able to: appreciate limitations of his/her genetics expertise, understand the social and psychological implications of genetic services, and know how and when to make a referral to a genetics professional.[57] The NCHPEG also recognizes the need for continuing medical education among geneticists.

[56] NCHPEG, *Core Competencies in Genetics Essential for All Health Care Professionals,* January 2005, accessed from www.nchpeg.org on October 12, 2006.
[57] Ibid.

GENETIC SERVICES IN TEXAS:
FINDINGS AND RECOMMENDATIONS

SUMMARY OF FINDINGS

- Traditionally, genetic services have focused on newborn screening, reproductive health, and birth defects. It is increasingly recognized that genes play a role in the development of disease across the lifespan. This calls for the availability of genetic services throughout the lifespan, and the integration of genetic services across medical specialties.

- Texas has a limited supply of physician geneticists and genetic counselors in relation to its population. This is further complicated by its poor distribution of geneticists and other health care providers throughout the state. The limited capacity and disparity in physician supply impact patient access to care.

- Patient access to care is further impacted by the state's physical size and long distances between health care providers, lack of public transportation in non-metropolitan areas, limited insurance reimbursement rates, and the high number of uninsured individuals and families.

- Ongoing demographic changes in Texas continue to affect the health care needs and delivery of services in the state. Trends, such as a growing ethnic diversity, childbearing at older ages, and socioeconomic disparities must be considered in defining service delivery priorities.

- There is a need to improve data collection, integration and reporting capabilities related to utilization of services, access to care, prevalence of genetic disorders, and efficacy of services.

- There is a need to enhance public understanding of genetics and its impact on overall health.

- Primary care providers and other non-geneticist physician specialists must be trained, at minimum, to recognize indications for a genetic referral, know where to refer patients needing genetic services, and work in concert with genetics professionals to coordinate and provide comprehensive care to individuals and families.

- Genetics physicians and counselors must be trained on technological advances in predictive testing, management, and treatment of genetic disorders. This training should be ongoing.

RECOMMENDATIONS

The Interagency Council for Genetic Services respectfully submits the following recommendations for consideration.

1. Access – increase access to genetic services in rural and underserved areas of the state.
    b. Allocate funds to implement telemedicine technology in outlying areas of the state. Resources needed include appropriate facilities, equipment, and technical assistance in the identification of professional partners and contract negotiation.
    c. Increase utilization of genetic counselors and advance practice nurses in genetic service delivery by allowing direct billing for their services.
    d. Strengthen primary care provider capacity through education and training to provide primary level genetic services.
    e. Provide financial assistance to students interested in becoming a physician geneticist or genetic counselor.

2. Education and Training – allocate resources to develop and implement comprehensive genetics education and training at all levels, i.e., primary care and other non-geneticist physician specialists, nurses, medical and nursing students, physician assistants, physician geneticists, genetic counselors, and the general public.
    a. Identify, modify, as appropriate, and deliver existing education models.
    b. Collaborate with genetic service providers and professional education groups to develop web-based training opportunities for geneticist and non-geneticist health care providers, awarding appropriate continuing education credits per discipline. Information updates would be made to assure awareness of emerging issues and appropriate utilization of new genetic technologies.
    c. Utilize telemedicine as an education medium.
    d. Work with schools of public health, medicine, and nursing to promote the integration of genetics into professional study and practice.
    e. Work with lay and professional groups to modify existing or develop new language-appropriate education materials for consumers. Focus group test products and revise accordingly before release.

3. Access – promote access to family-centered, culturally, and linguistically appropriate genetic services.
    a. Develop and employ recruitment strategies to attract diversity in genetics training programs.
    b. Provide cultural competency information and training opportunities to genetic service providers.

4. Data – allocate funds to the IACGS to improve data collection and reporting capabilities about incidence of genetic disorders, and utilization and efficacy of genetic services for health resource planning and improvement.

5. Safety Net Programs – increase funding allocation to address contract ceiling issues and to assist with plans for comprehensive education and training.

6. Policy – increase efforts to assure adequate and timely reimbursement for genetic services.