Interagency Council for Genetic Services

Resource Allocation Plan
2010-2011

The 2010-2011 Resource Allocation Plan is Prepared by the Interagency Council for Genetic Services
In Compliance with Human Resource Code
Title 9, Chapter 134, Section 134.0041
71st Texas Legislature

November 2008
2010-2011 Resource Allocation Plan

Table of Contents

Preface ................................................................................................................................. 1

Executive Summary ............................................................................................................ 2

Introduction ...................................................................................................................................6

What are genetic disorders? .......................................................................................... 6
What are genetic services? ............................................................................................. 7
What are the benefits of genetic services? ..................................................................... 8
Who provides genetic services? .................................................................................... 9

Genetic Services in Texas: An Inventory of Resources.........................................................11

Clinical Genetic Service Providers .............................................................................. 11
Genetics Clinics ............................................................................................................. 11
Genetics Laboratories .................................................................................................. 13

Population-Based Programs .......................................................................................... 13

Newborn Screening Program ............................................................................................ 13
Texas Early Hearing Detection and Intervention Program .............................................. 15
Texas Birth Defects Registry ............................................................................................ 16
Texas Teratogen Information Service .............................................................................. 16

Rehabilitative and Intervention Programs ........................................................................17

Department of Assistive and Rehabilitative Services ..................................................... 17
Department of State Health Services .............................................................................. 18
Department of Aging and Disability Services .................................................................. 19

Funding Sources ..............................................................................................................21
Medicaid .................................................................................................................. 21
Children’s Health Insurance Program ................................................................. 23
Title V Genetic Services ....................................................................................... 24
Newborn Screening Benefits Program ................................................................. 25
Private Health Care Coverage ............................................................................. 25
No Health Care Coverage .................................................................................... 25

Genetic Services in Texas: Challenges and Opportunities .................................. 27
Genetic Services in Texas: Findings and Recommendations ........................... 31

List of Tables

Table 1. Indications for a Genetic Referral by Stage of Life ................................... 10

Table 2. Number and Percentage of Newborns Screened, Confirmed
And Treated ........................................................................................................... 14

Table 3. Births to Texas Women ≥35, 2000-2003 ............................................... 28

List of Figures

Figure 1. Title V Genetic Clinic Sites in Texas ..................................................... 12

Figure 2. Percentage of Population without Health Insurance,
U.S. and Texas, 2005 ......................................................................................... 26
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 2010-2011 Resource Allocation Plan may be directed to:

Mary K. Kukolich, M.D., Chair
Interagency Council for Genetic Services
c/o Texas Department of State Health Services
1100 West 49th Street, MC 1918
Austin, Texas 78756
(512) 458-7111, extension 6675
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:

- 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,
- High rate of uninsured individuals and families, and
- Increased rate of women seeking emergency prenatal care and not having Medicaid.

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.
- Establish mechanisms for credentialing genetic counselors so that they may be eligible for reimbursement by Medicaid and the state.

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

1. **Structure of Interagency Council for Genetic Services** – The IACGS recommends that the council membership be restructured to reflect the reorganization of the health and human services agencies accomplished in 2003 through the passing of House Bill 2292. Human Resources Code, Chapter 134, establishes the IACGS and describes its membership as including representatives of Texas Department of Mental Health and Mental Retardation and Texas Department of Health, both agencies that were re-organized through HB 2292. It is also recommended that representation include the Department of Assistive and Rehabilitative Services (DARS), specifically from the Early Childhood Intervention Services (ECI). The IACGS also recommends adding representation from the genetic counselor professional organization of Texas.

2. **Duties of the Interagency Council for Genetic Services** – One of the requirements assigned to the council in 1987 when it was created was the biennial development of a resource allocation plan. Chapter 134, Section 134.0041 of the Human Resources Code further directs the council to “hold public hearings to gather information necessary to prepare the plan.” It is recommended that the holding of public hearings be reconsidered because the quarterly meetings of the council are public meetings. Comments provided during the public comment portion of IACGS meetings could be considered for inclusion in the resource allocation plan.

3. **Access** – 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.
   b. Increase utilization of advance practice nurses in genetic service delivery by allowing direct billing for their services.
c. Enable genetic counselors to direct bill.
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.
f. Establish means of reimbursing providers of genetic services for travel and costs associated with conducting outreach clinics.

4. **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.

5. **Cultural Competency** – 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.

6. **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; for members to travel to IACGS meetings; and for staff to support the council’s duties, which include surveying current resources for human genetic services in the state; initiating scientific evaluation of the current and future state needs for genetic services; and development of a data base for comparison of genetic services.

7. **Safety Net Programs** – increase funding allocation to address contract ceiling issues and to assist with plans for comprehensive education and training.
8. **Policy** – 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 50% of first trimester miscarriages are caused by chromosomal abnormalities.¹
- Birth defects / congenital anomalies is one of the leading causes of infant death in the United States.²
- In Texas, over 14,000 babies are born each year with one or more major structural malformation. The second leading cause of death among Texas infants is birth defects, accounting for almost 25% of infant deaths.³
- The expanded Newborn Screening Program identified 61 confirmed cases of metabolic disorders in 2007.⁴

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 an error in a single gene, missing or extra chromosome material, 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 disorders. A small number of these conditions result from mutations in mitochondrial 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 disorders, making their cumulative effect more significant. Sickle cell disease, cystic fibrosis, phenylketonuria (PKU), Marfan syndrome, and Huntington disease are examples of single-gene disorders. Symptoms range from mild to severe, may be present at birth or develop later in life, or may skip a generation depending upon the

---
³ Department of State Health Services (DHS), Birth Defects Epidemiology and Surveillance Branch, About Birth Defects in Texas, accessed from http://www.dshs.state.tx.us/birthdefects/about.shtm on September 10, 2008.
⁴ DHS, Newborn Screening Program data, calendar year 2007.
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 amount or structure. It is possible to inherit some types of chromosomal disorders, and it is possible for chromosomal abnormalities to occur in successive pregnancies. However, most of these disorders are not passed down from generation to generation. The errors most often 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 can be much less pronounced. Trisomy (extra chromosome), deletion (missing chromosome), and translocation (rearrangement of chromosomes) are the different types of chromosome problems.

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. 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, cancer, and diabetes.

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 twenty-seven disorders, many 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 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 a 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 have a significant risk of some type of heart or blood vessel problem. Typically, there is narrowing in the aorta or in the pulmonary arteries, ranging from trivial to severe and often requiring surgical correction of the defect.
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. Early detection and treatment can prevent or reduce disability and prevent long-term dependence on costly state services.

*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>
<thead>
<tr>
<th>Stage of Life</th>
<th>Indications</th>
</tr>
</thead>
</table>
| 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. The American Board of Medical Genetics lists 126 board certified geneticists in Texas; but only 8 providers in Texas bill for genetic services through the Title V Genetic Fee-for-Services Program. Some of these physicians do provide outreach to other regions of the state but outreach is limited. Relatively few geneticists must cover multiple sites. An additional concern related to the shortage of geneticists is that many of the limited pool are reaching retirement age.

GENETICS CLINICS
Figure 1 illustrates the types and locations of clinics providing Title V genetic services in Texas. There are a total of 31 clinics. 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 two part-time physician geneticists are housed. These physicians travel to Cook Children’s outreach clinics in Abilene, Amarillo, San Angelo, Waco, Denton, and Midland where they provide full services in limited frequency, e.g., one full-day clinic per month or quarter.

In Texas, there are 8 Title V Genetic Fee-for-Services 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. The 8 Title V contractors provide full services on a limited frequency at 23 outreach clinics throughout the state.
Figure 1. Title V Genetic Clinic Sites in Texas

Source: DSHS, Title V Genetic Fee-for-Services, FY 2009. REVD1108
Consistent with health care provider distribution, clinics are sparsely located throughout most of the state, with clusters of clinics in the Houston and Dallas-Fort Worth areas. Although there are other genetic clinics in major metropolitan areas that do not contract with Title V, families living in certain rural parts of the state must travel long distances to access genetic services. Genetests.org, a publicly funded medical genetics information website, is one source that may list other genetic services providers in Texas.

**GENETICS LABORATORIES**

Texas has genetic laboratories that are either university-based, private, or state-operated. The laboratories provide genetic testing and concentrate on one or more of four areas of service: cytogenetic (chromosomal abnormalities), biochemical (inborn errors of metabolism), mitochondrial (cellular energy), and/or DNA diagnostic (gene mutations).

**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). All three are supported by state and federal funds. One other population-based program is funded through the Title V funds and is operated from the University of North Texas.

- **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 began screening all Texas newborns for 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 2 shows the number of newborns screened at least once, results, and follow-up for Calendar Year 2007. A total of 16,192 babies were identified with presumptive positive screens and referred for confirmatory testing. A total of 432 were confirmed as having a genetic disorder, all of whom received treatment.
<table>
<thead>
<tr>
<th>Type of Screening Tests</th>
<th>Receiving at least one Screen(1)</th>
<th>No. of Presumptive Positive Screens</th>
<th>No. Confirmed Cases(2)</th>
<th>No. Needing Treatment Who Received Treatment(3)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>No.</td>
<td>%</td>
<td></td>
<td>No.</td>
</tr>
<tr>
<td>Phenylketonuria</td>
<td>396,000</td>
<td>95.6</td>
<td>126</td>
<td>14</td>
</tr>
<tr>
<td>Congenital Hypothyroidism</td>
<td>396,000</td>
<td>95.6</td>
<td>7,893</td>
<td>188</td>
</tr>
<tr>
<td>Galactosemia</td>
<td>396,000</td>
<td>95.6</td>
<td>646</td>
<td>6</td>
</tr>
<tr>
<td>Sickle Cell Disease</td>
<td>396,000</td>
<td>95.6</td>
<td>489</td>
<td>160</td>
</tr>
<tr>
<td>Other Screening</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Biotinidase Deficiency</td>
<td>396,000</td>
<td>95.6</td>
<td>129</td>
<td>12</td>
</tr>
<tr>
<td>Homocystinuria</td>
<td>396,000</td>
<td>95.6</td>
<td>189</td>
<td>2</td>
</tr>
<tr>
<td>Tyrosinemia</td>
<td>396,000</td>
<td>95.6</td>
<td>215</td>
<td>0</td>
</tr>
<tr>
<td>Congenital Adrenal Hyperplasia (CAH)</td>
<td>396,000</td>
<td>95.6</td>
<td>4,501</td>
<td>23</td>
</tr>
<tr>
<td>Maple Syrup Urine Disease (MSUD)</td>
<td>396,000</td>
<td>95.6</td>
<td>68</td>
<td>0</td>
</tr>
<tr>
<td>Medium Chain AcylCo-A Dehydrogenase (MCAD)</td>
<td>396,000</td>
<td>95.6</td>
<td>131</td>
<td>11</td>
</tr>
<tr>
<td>Beta-ketothiolase deficiency</td>
<td>396,000</td>
<td>95.6</td>
<td>336</td>
<td>0</td>
</tr>
<tr>
<td>Isovaleric Acidemia</td>
<td>396,000</td>
<td>95.6</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Methylmalonic Acidemia</td>
<td>396,000</td>
<td>95.6</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td>Propionic Acidemia</td>
<td>396,000</td>
<td>95.6</td>
<td>277</td>
<td>0</td>
</tr>
<tr>
<td>Carnitine Uptake Defect</td>
<td>396,000</td>
<td>95.6</td>
<td>210</td>
<td>0</td>
</tr>
<tr>
<td>3-Methylcrotonyl-CoA Dehydrogenase Deficiency</td>
<td>396,000</td>
<td>95.6</td>
<td>336</td>
<td>3</td>
</tr>
<tr>
<td>Long-Chain Hydroxyacyl-CoA Dehydrogenase Deficiency</td>
<td>396,000</td>
<td>95.6</td>
<td>242</td>
<td>1</td>
</tr>
<tr>
<td>Trifunctional Protein Deficiency</td>
<td>396,000</td>
<td>95.6</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Very-Long-Chain Acyl-CoA Dehydrogenase Deficiency</td>
<td>396,000</td>
<td>95.6</td>
<td>67</td>
<td>4</td>
</tr>
<tr>
<td>Glutaric Acidemia Type I</td>
<td>396,000</td>
<td>95.6</td>
<td>336</td>
<td>7</td>
</tr>
<tr>
<td>Hydroxymethylglutaric Aciduria</td>
<td>396,000</td>
<td>95.6</td>
<td>0</td>
<td>0</td>
</tr>
</tbody>
</table>

(1) Use occurrent births as denominator.
(2) Report only those from resident births.
(3) Use number of confirmed cases as denominator.

---

5 DSHS, Newborn Screening Program data, Calendar Year 2007.
Note that some disorders are grouped on the chart. For example, the category Sickle Cell Disease includes Sickle Cell Anemia, Sickle Beta Thalassemia, and Sickle-Hemoglobin C Disease. A complete list of the disorders screened by DSHS can be found at: [http://www.dshs.state.tx.us/newborn/quickreference.shtm](http://www.dshs.state.tx.us/newborn/quickreference.shtm).

- **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 discharge from a birthing hospital. 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.\(^6\)\(^7\)

  TEHDI provides oversight to hospitals and birth centers that screen newborns for hearing loss and report data to the state. In calendar year 2007, 377,462 newborns were screened for hearing loss. Just over 10,045 babies were referred for follow-up screening and evaluation. Statistically, about 1,000 will receive a confirmed diagnosis of moderate to profound hearing loss.\(^8\)

  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.\(^9\) 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.\(^10\)\(^11\)

---


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. In addition to TEHDI, DSHS administers the Vision and Hearing Screening Program, a 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 five-year period 1999 to 2004, the Registry reported a total of 81,753 infants and fetuses with any monitored birth defect, a prevalence of 370.13 per 10,000 live births. 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 miscarriage and 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 TTIS is a statewide counseling and information program for individuals who have questions or concerns about exposures to teratogens during pregnancy. TTIS also serves as a resource and distributes information to health care providers. The TTIS has a toll free Pregnancy Riskline (800-733-4727) for patients and health care providers to call to receive counseling and information. The staff at TTIS also travel throughout Texas to give presentations to both the public and 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 250% of the Federal Poverty Income Level do not pay for services.14 Others pay a cost share for services based on a sliding fee scale.

In FY 2007, ECI provided comprehensive and follow along services to more than 49,359 children through its provider network at school districts, regional

---

education service centers, community mental health and mental retardation centers, and private nonprofit organizations.\textsuperscript{15}

- **Deaf and Hard of Hearing Services** – The DARS Office for Deaf and Hard of Hearing Services is available for persons of all ages who are deaf or hard of hearing. The goal of services is to eliminate societal and communication barriers to improve equal access for people who are deaf or hard of hearing.\textsuperscript{16}

- **Services for the Blind and Visually Impaired** – The Division for Blind Services assists blind or visually impaired people and their families in regaining independence and finding jobs. The programs include: Vocational Rehabilitation, Independent Living Rehabilitation, and the Transition Program. The Criss Cole Rehabilitation Center offers intensive vocational and independent living training in a residential program in Austin, Texas.\textsuperscript{17}

- **Services for Persons with Physical and Mental Disabilities** – The DARS Division for Rehabilitation Services is the state’s principal authority on the vocational rehabilitation of Texans with disabilities. Programs include the Vocational Rehabilitation program; Transition Planning; and Supported Employment. Independent living services are also offered including centers that promote self-sufficiency.\textsuperscript{18}

**Department of State Health Services (DSHS)**

- **Children with Special Health Care Needs (CSHCN) Services Program** – The program provides health benefits to individuals younger than 21 who have a chronic physical or developmental condition, and to individuals of any age who have cystic fibrosis. The health care benefits provided include payments for medical care, family support services, and related services not covered by Medicaid, CHIP, private insurance, or other “third party payors.” The program also contracts with community-based organizations throughout the state to provide clinical and support services to children with special health care needs and their families. The services covered by the program health benefits include: primary and preventive care, speech and hearing services, vision care, mental health services, ambulance, hospital care, hospice care, medicines, special

\textsuperscript{15} DARS, *ECI Fact Sheet*, accessed from [www.dars.state.tx.us/ecis/factsheet.shtml](http://www.dars.state.tx.us/ecis/factsheet.shtml) on September 30, 2008.

\textsuperscript{16} Department of Assistive and Rehabilitative Services (DARS), *Services for Persons who are Deaf or Hard of Hearing*, accessed from [www.dars.state.tx.us/dhhs.index.shtml](http://www.dars.state.tx.us/dhhs.index.shtml) on September 30, 2008.

\textsuperscript{17} DARS, Services for the Blind and Visually Impaired, accessed from [www.dars.state.tx.us/dbs/index.shtml](http://www.dars.state.tx.us/dbs/index.shtml) on September 30, 2008.

\textsuperscript{18} DARS, *Services for Persons with Physical and Mental Disabilities*, accessed from [www.dars.state.tx.us/drs.index.shtml](http://www.dars.state.tx.us/drs.index.shtml) on September 30, 2008.
nutritional products and services, care by medical specialists, home health nursing, physical and occupational therapy, meals, lodging, and transportation when needed to obtain medical care, and other support services. Medicaid, CHIP, and commercial health insurance benefits, if any, must be used before using CSHCN health benefits.19

- **Program for Amplification for Children of Texas (PACT)** – 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.20 PACT provides about 2,935 hearing aids each year to Texas children.21

**Department of Aging and Disability Services (DADS)**

DADS services are designed to enable older and disabled Texans to live dignified, independent, and productive lives in a safe living environment. Services include in-home services, community-based services, and institutional services for people who require that level of support. Several DADS services are designed for persons with Mental Retardation (MR) and Related Conditions (RC). In most cases, the disability is present from birth, and in many cases, of genetic origin. DADS data contain limited information about diagnoses, and it is not possible to identify how many persons with genetic conditions are served by DADS.

- **Medically Dependent Children Program (MDCP)** – provides services for families caring for children who are medically dependent on technology as an alternative to residing in a nursing facility. Services include adaptive aids, minor home modifications, respite, and transition assistance services. MDCP served an average of 1,508 persons per month in FY 2007. As of June 2008, the Interest List for MDCP services included 9,920 individuals, with an average time on the list of 1.1 years.

- **Community Living Assistance and Support Services (CLASS)** - provides services for individuals with related conditions as an alternative to residing in an Intermediate Care Facility for Persons with Mental Retardation (ICF/MR). Services include adaptive aids and medical supplies, case management, minor

---

home modifications, nursing services, occupational and physical therapy, and transition assistance. In FY 2007, the program served an average of 3,113 persons per month. As of June 2008, the Interest List for CLASS services included 21,496 individuals, with an average time on the list of 2.4 years.

- **Home and Community-Based Services (HCS)** - provides services for individuals with mental retardation or a related condition as an alternative to residing in an ICF/MR. Services include case management, residential assistance, supported employment, adaptive aids, minor home modifications, and/or specialized therapies such as occupational therapy, physical therapy, audiology, speech/language pathology, and licensed nursing services. Over 11,000 persons were served per month during FY 2007. The Interest List for HCS services as of June 2008 included 37,187 individuals, with an average time on the list of 3.5 years.

- **Deaf Blind Multiple Disabilities (DBMD)** - provides services for individuals with deaf blindness and one or more other disabilities as an alternative to residing in an ICF/MR. Services include adaptive aids and medical supplies, dental services, assisted living, minor home modifications, nursing services, physical therapy, dietary services, and transition assistance.

- **Texas Home Living Waiver (TxHmL)** - provides essential services and supports for individuals with mental retardation or a related condition as an alternative to residing in an ICF/MR. Services include community support, employment assistance, skilled nursing, behavioral support, minor home modifications, dental treatment, and specialized therapies. Coordination of services is provided by the local mental retardation authority service coordinator.

- **Intermediate Care Facilities – Mental Retardation (ICFs/MR)** – serve four or more individuals with mental retardation or related conditions. ICFs/MR are operated by both private and public (community MHMR centers and state schools) entities. These facilities provide diagnosis, treatment, rehabilitation, ongoing evaluation, planning, 24-hour supervision, coordination, and integration of health or rehabilitative services.

- **Mental Retardation State Schools Services** - Texas has eleven state school campuses and two state centers providing mental retardation residential services. State schools and centers provide 24-hour residential services, comprehensive behavioral treatment services and health care services as well as skills training, vocational programs, and services to maintain connections between residents and their natural support systems. Approximately 5,000 persons reside at the state schools.

- **Mental Retardation Community Services** - services are provided to persons with mental retardation who reside in the community. These services do not include services provided through ICF/MR facilities and Medicaid waiver
programs. MR Community Services assist individuals to participate in age-appropriate community activities and services. These services include assessment, community supports, employment services, respite, and therapies.22

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 that neither the federal nor state government can limit the number of eligible people who can enroll and receive services covered under the program. In July 2006, approximately one in nine Texans relied on Medicaid. HHSC reported the unduplicated number of Texans receiving Medicaid in State Fiscal Year 2005 as 3.7 million at an estimated cost of $17.9 billion.23

Medicaid covers three primary groups:

1. Families and children – based on income level and depending on age or pregnancy status. Families and children represent 68% of the total Medicaid caseload, the largest share of Medicaid clients.

2. Cash assistance recipients – based on receipt of Temporary Assistance to Needy Families (TANF) or Supplemental Security Income (SSI).

3. Aged and disabled – based on income level, age, and physical or mental disability.24

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 organization (HMO) or primary care case management (PCCM). HMOs are licensed by

22 Department of Aging and Disability Services (DADS), Overview of DADS Long-Term Services and Supports, accessed from agency staff, October 2, 2008.
24 Ibid.
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. 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.

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.27

DSHS serves as a resource for Medicaid policy development for genetic services. Providers enrolled as Medicaid approved genetic service providers must meet certain requirements. The provider’s medical director must be a clinical geneticist (doctor of medicine [MD] or doctor of osteopathy [DO]) who is licensed by the Texas Medical Board and who is board eligible / certified by the American Board of Medical Geneticists (ABMG). Prior to enrollment, application qualifications for the provision of genetic services are verified and approved by DSHS. The laboratory used for confirmatory testing must comply with Clinical Laboratory Improvement Amendments (CLIA) rules and regulations. Genetic providers are reimbursed according to an established allowable maximum fee schedule. The services that are reimbursable are evaluation, diagnosis, counseling, and follow-up of clients with known or suspected genetic disorders.28

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. 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

27 Ibid.
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 (that is, one showing no physical abnormalities or an indication of a problem). In order to determine if there is a problem, both parents should receive chromosome testing. Medicaid will pay for the mother to be tested, but will not cover the father.

In the case study above, the father 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 a semi-annual enrollment fee that covers all the children in the family. Families must also pay co-payments. Both enrollment fees and co-pays are based on the family income. Enrollment fees do not exceed $50 for each six-month term of eligibility; co-payments for doctor visits and prescription medicines range from $3 to $20.²⁹

To qualify for CHIP, a child must be under age 19, a Texas resident, and a U.S. citizen or legal permanent resident. In addition, the child must be living in a family whose income is at or below 200 percent of the Federal Poverty Level (FPL). Services are delivered by private managed care organizations (MCO). Because of the limited number of clinical geneticists in Texas or because long distance travel is a barrier, a patient may need to see an out-of-network provider. In these cases, reimbursement for claims is at the discretion of the MCO. Some genetic service providers have reported difficulty in receiving payment, either having the claim rejected or only receiving partial payment.

---

In 2005, the 79th Legislature authorized HHSC to establish the CHIP Perinatal Program, a program designed to provide health benefit coverage to unborn children. Rolled out in January 2007, the CHIP Perinatal program provides prenatal care to the unborn children of women who do not qualify for Medicaid, including legal permanent immigrants and undocumented immigrants.30

**Title V Genetic Services**

Title V of the Social Security Act provides 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.

Fiscal Year 2009 Title V funds allocated to genetic services total just under $900,000. A portion of that stipend is allocated to a population-based education program. In comparison, FY 1997 dollars allocated to Title V genetic services were $1,834,134. Some 3,892 unduplicated clients received clinical genetic services through Title V in FY 2007, with another 769 people reached through the population-based Texas Teratogen Information Service. In FY 2009, there are a total of seven contracted providers of clinical genetic services and two population-based contracted programs. The clinical service providers are located in: Austin, Corpus Christi, El Paso, Fort Worth, Galveston, Houston, and McAllen.

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

---

30 Ibid.
generation of third-party user fees. 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.

Newborn Screening Benefits Program
Following the expansion of the newborn screening panel, the Department of State Health Services instituted the Newborn Screening (NBS) Benefits Program. The NBS Benefits Program assists Texas families who do not have insurance or another funding source to pay for medically necessary services. The program was developed to ensure the availability of confirmatory testing, evaluation, and management of inheritable disorders, detected by the Department of State Health Services (DSHS) NBS Program. NBS Benefits Open Enrollment was initiated to procure these clinical services through contracts with public and private providers. The NBS Program will reimburse NBS Benefits Contractors for providing specified services to eligible clients, as funding allows. Eligible clients may receive confirmatory testing, evaluation, vitamins, formula, medication, and follow-up care as needed at no cost or at a reduced cost. To receive the services, clients must have a confirmed diagnosis of a disorder screened by the NBS Program, and continue to meet the eligibility criteria.

Private Health Care Coverage
In 2005, 55% of non-elderly Texans received health care coverage through employer-based private health insurance. In 2004, 19 percent of U.S. working adults under the age of 65 were uninsured. In comparison, 28 percent of working adult Texans were uninsured. These uninsured adults either work in jobs that do not provide health insurance or are unable to afford the coverage offered.

No Health Care Coverage
According to the U.S. Census Bureau, the two-year average percentage of people without health insurance coverage for 2006 and 2007 was 15.5 percent for the U.S. and 24.5 percent for Texas. While there is variability among Texas counties, every major

33 Ibid.
Texas city has an uninsured rate that is higher than the national average.\textsuperscript{35} In a comparison of three-year average uninsured rates for 2005-2007, Texas led all states in the percentage of uninsured people.\textsuperscript{36} Figure 2 compares uninsured rates in the United States and Texas.

\begin{figure}[h]
\centering
\includegraphics[width=0.5\textwidth]{chart}
\caption{People without Health Insurance Coverage, All Ages Two-Year Average, 2006-2007}
\end{figure}


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

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.

\textsuperscript{35} Texas Comptroller of Public Accounts, \textit{The Uninsured: A Hidden Burden on Texas Employers and Communities}, April 2005, accessed from \url{www.window.state.tx.us/specialrpt/uninsured05/} on September 11, 2006.

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. Individuals and families needing genetic services often must travel great distances to see a genetic specialist.

While many geneticists provide periodic clinics in outlying areas, these occur in limited frequency. Patients 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. Often the cost of travel is paid for through Medicaid funds. This is further complicated by limited public transportation outside of metropolitan areas. The majority of Texas counties are rural or non-metropolitan.

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. In 2026, the Hispanic population 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 in poverty. In 2007, 16.5% of Texans were living at or below the Federal Poverty Income Level.

---

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.40

<table>
<thead>
<tr>
<th>Table 3: Births for the State of Texas</th>
</tr>
</thead>
<tbody>
<tr>
<td>Age Group: 35 plus</td>
</tr>
<tr>
<td><strong>Year</strong></td>
</tr>
<tr>
<td>Births</td>
</tr>
<tr>
<td><strong>Percent Increase in Births to Age Group 35 Plus</strong></td>
</tr>
</tbody>
</table>

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.41 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 less than $900,000. This funds eight fee-for-service providers and two population-based

---

40 Department of State Health Services, Center for Health Statistics, 2008.
programs, 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’ (IACGS) 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.

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.\(^{42}\)

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 materials and educational programs have been and continue to be developed.\(^{43}\) 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.\(^{44}\) The NCHPEG also recognizes the need for continuing medical education among geneticists.


\(^{44}\) 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. Structure of Interagency Council for Genetic Services – The IACGS recommends that the council membership be restructured to reflect the re-organization of the health and human services agencies accomplished in 2003 through the passing of House Bill 2292. Human Resources Code, Chapter 134, establishes the IACGS and describes its membership as including representatives of Texas Department of Mental Health and Mental Retardation and Texas Department of Health, both agencies that were re-organized through House Bill 2292. It is also recommended that representation include the Department of Assistive and Rehabilitative Services (DARS), specifically from the Early Childhood Intervention Services (ECI). The IACGS also recommends adding representation from the genetic counselor professional organization of Texas.

2. Duties of the Interagency Council for Genetic Services – One of the requirements assigned to the council in 1987 when it was created was the biennial development of a resource allocation plan. Chapter 134, Section 134.0041 of the Human Resources Code further directs the council to “hold public hearings to gather information necessary to prepare the plan.” It is recommended that the holding of public hearings be reconsidered because the quarterly meetings of the council are public meetings. Comments provided during the public comment portion of council meetings could be considered for inclusion in the resource allocation plan.

3. Access – 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.
   b. Increase utilization of advance practice nurses in genetic service delivery by allowing direct billing for their services.
   c. Enable genetic counselors to direct bill.
   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.
f. Establish means of reimbursing providers of genetic services for travel and costs associated with conducting outreach clinics.

4. **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.

5. **Cultural Competency** – 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.

6. **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; for members to travel to IACGS meetings; and for staff to support the council’s duties, which include surveying current resources for human genetic services in the state; initiating scientific evaluation of the current and future state needs for genetic services; and development of a data base for comparison of genetic services.

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

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