



# Interagency Council for Genetic Services

## Resource Allocation Plan 2012-2013

The 2012-2013 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  
71<sup>st</sup> Texas Legislature

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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. In 1989, Human Resources Code, Title 9, Chapter 134, Section 134.0041 was added by Acts 1989, 71<sup>st</sup> Legislature to require the IACGS to 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.

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.

Questions regarding the 2012-2013 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 detected during pregnancy and at 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 reimbursements,
- Highest rate of uninsured of all the states
- Decreasing rates for women receiving prenatal care. Between 2004 and 2008 the percentage of women delivering with no prenatal care has increased from 1.9% to 4.8%. Additionally, between 2004 and 2008 the percentage of women receiving the prenatal care during the 1<sup>st</sup> trimester has decreased from 81.8% to 59.0%.<sup>1</sup>

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, and know where to refer patients
- Access genetic services, and to 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.
- Assure funding so that providers of genetics services can function and provide outreach services.

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** – Restructure the council membership to reflect the re-organization of the health and human services agencies that was accomplished in 2003 by the passing of House Bill 2292. The Human Resources Code, Chapter 134, established the IACGS and describes its membership to including representatives of Texas Department of Mental Health and Mental Retardation and Texas Department of Health. Since both these agencies were re-organized through HB 2292, it is recommended IACGS representation reflect these changes by including a representative from the Department of Assistive and Rehabilitative Services (DARS) and specifically that representative be from the area of Early Childhood Intervention Services (ECI).
2. **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 would include: appropriate facilities and equipment, and technical assistance for identifying professional partners and negotiating contracts.
  - 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 a means of reimbursing providers of genetic services for travel and costs associated with conducting outreach clinics.
3. **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, and review effectiveness of current educational 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.

- c. Award appropriate continuing education credits for different disciplines.
  - d. Provide information updates to assure awareness of new emerging issues and to increase appropriate utilization of new genetic technologies.
  - e. Utilize telemedicine as an educational medium.
  - f. Work with schools of public health, medicine, and nursing to promote the integration of genetics into professional study and practice.
  - g. Work with lay and professional groups to modify existing or develop new language-appropriate education materials for consumers.
  - h. Utilize focus groups to test new education materials before their release.
4. **Cultural Competency** – Promote access to family-centered genetics services that are culturally and linguistically appropriate.
- a. Develop recruitment strategies to increase cultural diversity in genetics training programs.
  - b. Increase cultural competency of genetics service providers by providing culturally sensitive training opportunities.
5. **Data** – Improve the utilization and efficacy of genetic services health resource planning by increasing the allocation of funds to the IACGS in order to increase reporting capabilities and data collection regarding the incidence of genetic disorders. Increased funds would:
- a. Assist members to travel to IACGS meetings to support the council’s duties.
  - b. Assist members in surveying current resources for human genetic services in the state.
  - c. Allow for the initiation of scientific evaluation of the current and future state needs for genetic services.
  - d. Enhance the ability to develop a data base for comparison of genetic services.
6. **Safety Net Programs** – Increase funding allocation to address contract ceiling issues and to assist with plans for comprehensive education and training.
7. **Policy** – Increase efforts to assure adequate and timely reimbursement for genetic services.

## INTRODUCTION

Genetic disorders occur throughout the lifespan. Historically, there has been heavy utilization of genetic services during pregnancy and childhood. Consider the following facts:

- More than 50% of first trimester miscarriages are caused by chromosomal abnormalities in the embryo and approximately 1 in 150 babies born in the United States is born with a chromosomal abnormality.<sup>2</sup>
- According to the National Vital Statistics Reports, congenital malformations, deformations and chromosomal abnormalities are the number one in the top 10 causes of infant mortality in the United States for 2010.<sup>3</sup>
- The CDC estimates in the United States, birth defects are responsible for at least 30% of all pediatric hospital admissions and they are one of the leading causes of infant deaths.<sup>4</sup>
- In Texas, there were 19,697 infants or fetuses with any monitored birth defect in our Registry delivered in 2009.<sup>5</sup>
- Also in Texas in 2007, birth defects resulted in nearly 35,000 hospitalizations among infants in Texas and, with total charges of nearly \$2.4 billion based on hospital discharge data. Birth defects are the leading cause of death among infants in Texas. Due to the large population of Texas relative to other states, the total cost of hospitalization for infants with birth defects is high. At least half of all babies born with birth defects in Texas have more than one major malformation.<sup>6</sup>
- The expanded Newborn Screening Program identified 680 confirmed cases of disorders in 2009.<sup>7</sup>

### 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 pattern of inheritance. More severe effects include physical abnormalities, organ damage, intellectual disability and developmental disabilities, 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, or during embryonic development. Effects can be profound, including miscarriage, stillbirth, and if the newborn survives, severe physical deformity, damage to multiple organs, intellectual disability and developmental disability, and premature death, often within the first year of life. Other cases can be much less pronounced. Trisomy (extra chromosome), deletion (missing chromosome material), and translocation (rearrangement of chromosomes) are the different types of chromosome problems. With the development of microarray testing many new chromosomes disorders are being recognized and require complex genetic counseling services.

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 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-nine disorders, many of which may be treated through a combination of dietary supplements, medical formula, medications, vitamins and continued monitoring of the condition through blood, urine and other tests. Babies born in Texas are also screened for hearing loss. 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. Pre-symptomatic testing is available for individuals 18 years of age or older who are at risk for carrying the HD gene.

**Anticipatory care** for individuals with genetic conditions. 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, or she may have the option of preventative surgery.

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, physicians, physician assistants, nurse practitioners, clinical nurse specialists, and genetic laboratories. Currently, genetic counselors cannot provide billable services independently. 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, , and health educators all 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**

<p>Preconception and Prenatal</p>	<ul style="list-style-type: none"> <li>▪ Maternal age <math>\geq 35</math> – associated with higher incidence of preterm and low birth weight babies; maternal complications, such as gestational diabetes and hypertension; problems associated with chromosomal disorders</li> <li>▪ Maternal age <math>\leq 18</math> – associated with increased incidence of preterm and low birth weight babies</li> <li>▪ Family history of genetic disorder</li> <li>▪ Previous child with birth defect, chromosomal abnormality or other genetic disorder</li> <li>▪ Birth defect or sign of genetic condition identified on ultrasound</li> <li>▪ Abnormal serum screen for neural tube defects and chromosomal abnormalities</li> <li>▪ History of miscarriage or stillbirth</li> <li>▪ Exposure to infection, drugs, chemicals, toxins during pregnancy</li> <li>▪ Carrier testing for specific ethnic groups who have a higher incidence of genetic disorders, e.g., sickle cell disease, Tay-Sachs disease</li> </ul>
<p>Pediatric</p>	<p>Newborns, infants, children with</p> <ul style="list-style-type: none"> <li>▪ Birth defects or multiple congenital anomalies</li> <li>▪ Known or suspected genetic disorders, such as Down syndrome, cystic fibrosis, muscular dystrophy</li> <li>▪ Abnormal growth patterns, i.e., excessive growth, very short stature</li> <li>▪ Abnormal body and limb proportions</li> <li>▪ Ambiguous genitalia</li> <li>▪ Abnormal or unusual facial features</li> <li>▪ Developmental delays</li> <li>▪ Intellectual disability</li> <li>▪ Metabolic disorders</li> </ul>

Adult	<ul style="list-style-type: none"> <li>▪ Abnormal growth patterns, i.e., excessively tall or short</li> <li>▪ Adult onset genetic disorders, such as Huntington disease, Spinal Muscular Atrophy, Myotonic dystrophy</li> <li>▪ Positive history of familial disorders, e.g., breast, ovarian and colon cancers, Duchenne’s muscular dystrophy, Thalassemia, Tay-Sachs, sickle cell disease</li> </ul>
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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**

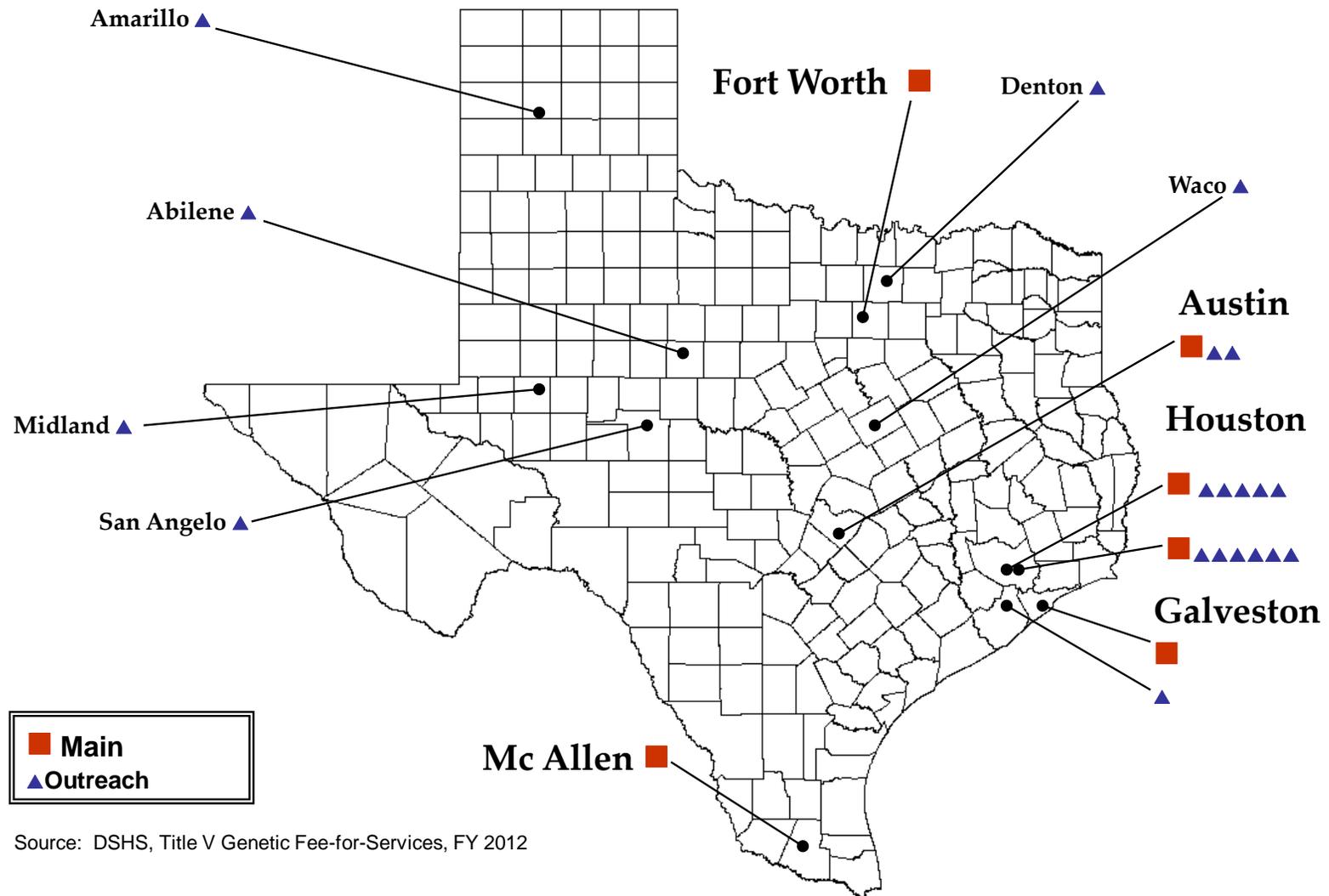
Clinical genetic services are medical services provided to individuals, families, and populations who have or are at risk for disorders with genetic implications. Services include testing, counseling, education, treatment, and where appropriate, referral for other services. Clinical genetic services 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 72 board certified clinical geneticists in Texas, in which 21 practice in one institution, and many of these physicians primary focus is research with very limited clinical involvement and others are not currently providing direct services. There are 6 institutions with 31 clinical geneticists (21 are located at the same institution) who 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 in Texas 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 26 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.

In Texas, there are 6 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 Austin. The 6 Title V contractors provide full services on a limited frequency at 20 outreach clinics throughout the state.

# Figure 1. Title V Genetic Clinic Sites in Texas



Source: DSHS, Title V Genetic Fee-for-Services, FY 2012

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, Title V supports population-based programs aimed at the early detection of disorders with a significant genetic association and referral to intervention services.

### **Texas Teratogen Information on Pregnancy Service Helpline (TIPS)**

TIPS is located at the University of Texas Health Science Center at Houston. In this program 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, etc.), substance (cigarette smoke, drugs, alcohol, etc.), infectious agent (rubella, toxoplasma, etc.) or maternal condition (diabetes, maternal PKU, etc.) 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, intellectual disabilities, 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 TIPS is a statewide counseling and information program for individuals who have questions or concerns about exposures to teratogens during pregnancy. TIPS also serves as a resource and distributes information to health care providers. The TIPS has a toll free Helpline (855-884-7248) for patients and health care providers to call to receive counseling and information. The TIPS program staff also travel throughout Texas to give presentations to both the public and health care providers.

### 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. Starting December 2009, the Newborn Screening Program added cystic fibrosis to its screening panel bringing the total number of screened disorders to 28 (29, including hearing screening). In September of 2012, Texas Department of State Health Services is adding severe combined immunodeficiency or SCID to the list of diseases that all newborns in Texas will be screened for at birth for a total of 29 disorders (30, including hearing screening).

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 types of screening tests and the number of confirmed cases for Calendar Year 2009. A total of 680 were confirmed as having a disorder.

<b>TABLE 2. Newborn Screening Identified Disorder and Number of Confirmed Cases</b>	
Total Births by Occurrence: <u>401,599</u> Reporting Year: <b>FY 2009</b>	
Type of Screening Tests	No. Confirmed Cases
Phenylketonuria	16
Galactosemia (includes Duarte variant)	134
Hemoglobinopathies*	175
Biotinidase Deficiency	42
Homocystinuria	2
Congenital Hypothyroidism	210
Tyrosinemia Type II	0
Congenital Adrenal Hyperplasia (CAH) (includes non-classical)	50
Maple Syrup Urine Disease (MSUD)	1
Medium Chain AcylCo-A Dehydrogenase (MCAD)	21
Beta-ketothiolase deficiency	0
2-Methylbutrylglycinuria	0
Isovaleric Acidemia	2
Methylmalonic Acidemia (mutase deficiency form)	0
Propionic Acidemia	2
Carnitine Palmitoyl Transferase Deficiency	0
Carnitine Acylcarnitine Translocase Deficiency	0
Carnitine Uptake Defect	0
3-Methylcrotonyl-CoA Dehydrogenase	9

Deficiency	
Long-Chain Hydroxyacyl-CoA Dehydrogenase Deficiency	1
Trifunctional Protein Deficiency	0
Very-Long-Chain Acyl-CoA Dehydrogenase Deficiency	8
Glutaric Acidemia Type I	4
Hydroxymethylglutaric Aciduria	0
Multiple Carboxylase Deficiency	0
Argininosuccinic Acidemia	1
Citrullinemia	2
Cystic Fibrosis (CF) - started being screened for in December 2009	N/A
	<b>TOTAL</b>
	<b>680</b>

Note: Figures may change from previous or future generated reports due to updates in diagnosis received from medical specialists.\*Note that some disorders are grouped on the chart. For example, the category Hemoglobinopathies includes Sickle Cell Anemia, Sickle Beta Thalassemia, and Sickle-Hemoglobin C Disease.<sup>8</sup>

A complete list of the disorders screened by DSHS can be found at:

<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.

The TEHDI program provides oversight to hospitals and birth centers that screen newborns for hearing loss and report data to the state. In calendar year 2010, 376,972 newborns were screened for hearing loss. Just over 12,110 babies were referred for follow-up screening and evaluation. Statistically, about 1,200 will receive a confirmed diagnosis of moderate to profound hearing loss.<sup>9</sup>

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. 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.

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 the TEHDI Program, 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 births in Texas statewide and identifies cases of birth defects. For the period 2000 to 2009, the Registry reported a total of 160,878 infants and fetuses with any monitored birth defect, a prevalence of 417 per 10,000 live births. Researchers use Registry data to conduct epidemiological studies to find preventable causes of birth defects in Texas. In addition, parents of children identified through the Registry are informed of health and human services available through state and federal programs.<sup>10</sup>

## **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 is a statewide program for families with children, birth to three, with disabilities and developmental delays. 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, and assistive technology services, among others. Families with children enrolled in Medicaid or CHIP, or another public benefit program listed in the Family Cost Share brochure whose income is at or below 200% of the Federal Poverty Level, do not pay for any ECI services. Other families pay a [cost share](#) determined by a sliding fee scale based on family size and adjusted income after allowable deductions.<sup>11</sup> In FY 2011, ECI provided comprehensive services to more than 59,000 children through its provider network of community/state mental health and mental retardation centers, private nonprofit and local organizations, regional education service centers and local independent school districts.
- **Deaf and Hard of Hearing Services** –The DARS Office for Deaf and Hard of Hearing Services (DHHS) works in partnership with people who are deaf or hard of hearing to eliminate societal and communication barriers to improve equal access for people who are deaf or hard of hearing. DHHS advocates for people of all ages who are deaf or hard of hearing to

enable them to express their freedoms, participate in society to their individual potential, and reduce their isolation regardless of location, socioeconomic status, or degree of disability.

- **Services for the Blind and Visually Impaired** – The DARS Division for Blind Services (DBS) assists blind or visually impaired individuals and their families. Depending on their goals and needs, DBS offers services to help regain independence or find a job. These services include: Vocational Rehabilitation, Independent Living Rehabilitation Program, Blind Children’s Vocational Discovery and Development Program, Transition Program, and the Blindness Education, Screening and Treatment, Program. The Criss Cole Rehabilitation Center, located in Austin, is a residential treatment program that offers intensive vocational and independent living training to Texans who are blind.
- **Services for Persons with Physical and Mental Disabilities** – The DARS Division for Rehabilitation Services (DRS) is the state’s principal authority on the vocational rehabilitation of Texans with physical and mental disabilities. Programs include the Comprehensive Rehabilitation Services, Independent Living Services and Centers, Supported Employment, Transition Planning, and the Vocational Rehabilitation Program.
- **Services for Persons with Autism** – The DARS Autism Program champions excellence in the delivery of services for families of children with autism. Services are provided through grant contracts with local community agencies and organizations that provide applied behavioral analysis (ABA) and other positive behavior support strategies. The program helps improve the quality of life for children on the autism spectrum and their families. To be eligible for services under a DARS grant, a child must meet three criteria:
  - Be 3 through 8 years of age (children lose eligibility on their ninth birthday)
  - Have a diagnosis on the autism spectrum
  - Be a Texas resident

Services in the DARS Autism program are provided by six contractors around the state. Any Texas resident can apply for services at any of these provider's offices. You do not need to live in the service area to qualify for services; however, transportation and lodging costs are not covered by this program.<sup>12</sup>

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

**Children with Special Health Care Needs (CSHCN) Services Program –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 that is not solely a delay in intellectual development or solely a mental, behavioral, or emotional condition, and to individuals of any age who have cystic fibrosis. Applicants must also meet the financial criteria for program eligibility of less than 200% of the federal poverty level based on the family size, and be a Texas resident. 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, dental services, speech and hearing services, vision care, mental health services, ambulance, hospital care, hospice care, medicines, special

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. The CSHCN Services Program has a waiting list for services. Clients on the waiting list may be removed from the list to receive services as funds become available.<sup>13</sup>

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

DADS services are designed to enable individuals who are older and individuals with disabilities to live dignified, independent, and productive lives. Services include in-home services, community-based services, and institutional services for people who require that level of support. Several DADS services are targeted to individuals with intellectual and developmental disabilities. 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 the number of individuals with genetic conditions who are served by DADS. While the term intellectual disability is preferred, state and federal law often still use the term “mental retardation”.

- **Medically Dependent Children Program (MDCP)** – provides a variety of services to support families caring for children who are medically dependent, and to encourage de-institutionalization of children in nursing facilities. Services include adaptive aids, minor home modifications, respite, and transition assistance services. MDCP served an average of 2,437 individuals per month in FY 2011. As of March 2012, the interest list for MDCP services included 24,289 individuals.
- **Community Living Assistance and Support Services (CLASS)** - provides home and community-based services for people with related conditions as an alternative to residing in an intermediate care facility for individuals with an intellectual disability or related conditions (ICF/IID). Services include adaptive aids and medical supplies, case management, minor home modifications, nursing services, occupational and physical therapy, and transition assistance. In FY 2011, the program served an average of 4,623 individuals per month. As of March 2012, the interest list for CLASS services included 41,434 individuals.
- **Home and Community-Based Services (HCS)** – provides individualized services and supports to people with diagnoses of an intellectual disability or related conditions as an alternative to residing in an ICF/IID. 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. HCS served an average of 19,382 individuals per month in FY 2011. The interest list for HCS services as of March 2012 included 56,966 individuals.

- **Deaf Blind Multiple Disabilities (DBMD)** - provides home and community-based services to people who have deaf blindness with a third disability as an alternative to residing in an ICF/IID. The DBMD program focuses on increasing opportunities for individuals to communicate and interact with their environment. Services include adaptive aids and medical supplies, dental services, assisted living, minor home modifications, nursing services, physical therapy, dietary services, and transition assistance. The program served an average of 151 individuals per month in FY 2011. As of March 2012, there were 480 individuals on the interest list.
- **Texas Home Living Waiver (TxHML)** - provides services and supports for individuals with intellectual and developmental disabilities as an alternative to residing in an ICF/IID. 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. The program served an average of 901 individuals per month in FY 2011.
- **Intermediate Care Facilities – Intellectual Disability or Related Conditions (ICFs/IID)** – provides residential and habilitation services to individuals with a diagnosis of an intellectual disability or related conditions. ICFs/IID are operated by both private and public entities. These facilities provide diagnosis, treatment, rehabilitation, ongoing evaluation, planning, 24-hour supervision, coordination, and integration of health or rehabilitative services. ICFs/IID served an average of 5,612 individuals per month in FY2011.
- **State Supported Living Centers** - Texas has 13 state supported living centers (formerly called state schools and state centers) providing residential services to individuals with an intellectual disability and intense behavioral or medical needs. State supported living 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. In FY 2011, an average of 4,072 individuals per month resided in state supported living centers.
- **Local Authority Intellectual and Developmental Disability Community Services** - provides services to people with intellectual and developmental disabilities who reside in the community. These services do not include services provided through ICFs/IID or Medicaid waiver programs. Services include eligibility determination, service coordination, community support, employment assistance, supported employment, nursing, behavioral support, specialized therapies, vocational training, and day habilitation. Services were provided to an average of 12,433 individuals per month in FY 2011.<sup>14</sup>

## 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 is 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. Approximately one in eight Texans relied on Medicaid for health insurance or long-term services and supports during SFY 2009. HHSC reported the number of Texans receiving Medicaid in December 2009 at 3.2 million. The total dollars (state and federal) spent for Medicaid in Federal Fiscal Year 2009 was 24.5 billion dollars (including disproportionate share hospital, or DSH).<sup>15</sup>

### Medicaid Covers Three Primary Groups

1. **Families and children** – based on income level and depending on age or pregnancy status. In State Fiscal Year 2009, non-disabled children represented 61% 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 persons with disabilities** – based on income level, age, and physical or mental disability.

As of March 1, 2012, Texas Medicaid providers are reimbursed through either the traditional fee-for-service model or through a Medicaid managed care plan (i.e. health maintenance organization (HMO)). The Primary Care Case Management program (PCCM), that served primarily rural counties in Texas, was phased out effective March 1, 2012. PCCM was replaced with STAR managed care, which was expanded to serve Texas Medicaid clients in the former PCCM counties under a newly formed Medicaid Rural Service Area (Medicaid RSA).

HMOs are licensed by the Texas Department of Insurance and deliver services under a risk-based arrangement. Per a [contract](#) between HHSC and each HMO, these organizations receive a monthly capitation payment for each person enrolled based on an average projection of medical expenses for the typical patient. In a managed care model, each Medicaid client must select a primary care provider, or be assigned to one. The PCP must provide a referral for most other health care services, such as specialty physician care, before the Medicaid managed care health plan will pay for them. Managed care providers must enroll as a Medicaid provider, as well as be credentialed and contracted with the Medicaid health plan(s) in their area to receive reimbursement. The rates are negotiated between the plan and provider, and documented in a contract. Non-contracted providers may be paid an out-of-network fee if the services cannot be provided by an in-network provider, or in emergencies.

Because the DSHS laboratory is the “sole source” provider of newborn screening in the state of Texas, the Health and Human Services Commission directly reimburses DSHS via an interagency contract for newborn screening performed by the DSHS laboratory when conducted for newborn Medicaid recipients. Newborn screening performed by the DSHS laboratory is therefore “carved out” of Medicaid managed care.

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.

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 in fee-for-service Medicaid. The services that are reimbursable include evaluation, diagnosis, genetic risk assessment, genetic laboratory tests (including interpretation and evaluation of results), management, treatment, counseling, and follow-up of clients with known or suspected genetic disorders. These same services are available through Medicaid managed care, however, reimbursement fees are negotiated between the providers and health plans via contractual arrangements.

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 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.*

### **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 an annual enrollment fee that covers all the children in the family. Co-payments also apply to certain services such as office visits, brand name drugs, non-urgent ER visits, and inpatient hospitalization. Co-payment amounts were increased effective March 1, 2012. 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 \$35.

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 79<sup>th</sup> 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 12 months of coverage to the unborn children of non-Medicaid eligible women. This benefit allows pregnant women who are not eligible for Medicaid due to income or immigration status to receive prenatal care, and provides CHIP benefits to the child upon delivery for the remainder of the 12-month coverage period.

Again, because the DSHS laboratory is the "sole source" provider of newborn screening in the state of Texas, the Health and Human Services Commission directly reimburses DSHS via an interagency contract for CHIP newborn screening performed by the DSHS laboratory when conducted for newborn CHIP recipients. (The Medicaid and CHIP newborn screening interagency contracts are separate documents.) Newborn screening performed by the DSHS laboratory is therefore "carved out" from the CHIP health plan premiums.<sup>16</sup>

### **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 2011 Title V funds allocated to genetic services total just under \$888,988. A portion of that stipend was allocated to a population-based education program. In comparison, FY 1997 dollars allocated to Title V genetic services were \$1,834,134. During FY 2011, 2,078 unduplicated clients received clinical genetics services through Title V. In FY2011, there were a total of six contracted providers of clinical genetic services and two population-based contracted programs. The clinical service providers were located in: Austin, 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 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, low-protein diet foods, medication, and follow-up care as needed. 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. The table below illustrates the growth of the NBS Benefits program from FY 2009 to FY 2011. The program has grown from 20 to 375 clients.

<b>TABLE 3. Newborn Screening Benefits Program</b>			
<b>Type</b>	<b>Client Breakdown</b>		
	FY09	FY10	FY11
Children 0-2	3	49	86
Children 3-5	1	33	50
Children 6-21	1	69	144
Pregnant Women	0	0	0
Adults	15	42	84
Subtotal	20	193	364
Presumptive Eligibility	3 (0)	21 (13)	29 (11)
<b>Totals</b>	<b>20</b>	<b>206</b>	<b>375</b>

See End Note <sup>17</sup>

### **Private Health Care Coverage**

In 2010, more than 51% of non-elderly Texans received health care coverage through employment-based, private health benefit plans.<sup>18</sup>

Access to private health plan coverage does not guarantee that the plan will provide reimbursement for genetic testing or other genetic services. Texas statutes require that HMO plans cover newborn screenings to detect genetic disorders but there is no similar statutory requirement for plans issued by insurance companies. In addition, while many employment-based plans are fully insured by an HMO or insurance company, the majority of employment-based plans are self-insured by the employer or plan sponsor and are not subject to state insurance statutes due to federal preemption laws.<sup>19</sup>

### **No Health Care Coverage**

According to the U.S. Census Bureau, the two-year average percentage of people without health insurance coverage for 2009 and 2010 was 16.2 percent for the U.S. and 25 percent for Texas.<sup>20</sup> In addition, 26.9 percent of Texans under the age of 65 did not have any type of public or private health coverage at any time during 2010.<sup>21</sup> In a comparison of three-year average uninsured rates for 2008-2010, Texas led all states in the percentage of uninsured people.<sup>22</sup>

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 health 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 health 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, availability of health care professionals and providers after hours and on weekends for non-emergent care, cultural competency of providers, and affordability of health care services, with or without health insurance coverage.

## **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.<sup>23</sup> Individuals and families needing genetic services often must travel great distances to see a genetic specialist. While some 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. Travel to receive genetic services is further complicated by the fact that public transportation is limited outside of most of our metropolitan areas and the majority of Texas counties are rural or non-metropolitan. Often the cost of travel is paid for through Medicaid funds.

The demographic history of Texas has been one of growth. Texas' population has increased more rapidly (in percentage terms) than the population of the nation in every decade since Texas became a state (Figure 2.1). The decade of the 1990s was notable in several regards, however, with the State's population growing to 20,851,820 by 2000, an increase of 22.8 percent since 1990. This increase of 3,865,310 persons was the largest of any decade in Texas history and moved Texas past New York to become the nation's second largest state. Texas population increase was second only to that in California (California increased by 4.1 million persons in the 1990s) and the eighth largest in percentage terms among all states.<sup>24</sup>

In 2010, Non-Hispanic Whites comprised 45% of the total Texas population, Hispanics 38%, Non-Hispanic Blacks 11%, and Non-Hispanic Other 6%. In 2004, minority populations, collectively, surpassed the size of the non-Hispanic white population in Texas. It is projected that by 2015, the Hispanic population will surpass the Non-Hispanic White population as the largest racial/ethnic group in Texas. Also projected is that by 2026, the Hispanic population is projected to become the majority population in Texas.<sup>25</sup>

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.<sup>26</sup> In 2009, 17.3 percent of Texans were living at or below the Federal Poverty Income Level.<sup>27</sup>

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 3 illustrates, Texas has seen a steady increase in births to women in this age group.

<b>TABLE 4. Births for the State of Texas</b>								
<b>Births</b>								
<b>Column Variable: Infant's Year of Birth</b>								
<b>Age of Mom: 35-99+ Years</b>								
<b>Mom's Place of Residence: Texas</b>								
<b>Year</b>	<b>2003</b>	<b>2004</b>	<b>2005</b>	<b>2006</b>	<b>2007</b>	<b>2008</b>	<b>2009</b>	<b>2010*</b>
<b>Births</b>	40,339	41,452	43,651	45,429	47,000	47,366	48,111	47,892
<b>Year</b>	<b>2003-2004</b>	<b>2004-2005</b>	<b>2005-2006</b>	<b>2006-2007</b>	<b>2007-2008</b>	<b>2008-2009</b>	<b>2009-2010*</b>	
<b>Percent Increase in Births to Age Group 35 Plus</b>	2.8%	5.3%	4.1%	3.5%	0.8%	1.6%	-0.5%	

\*2010 birth data are currently preliminary and subject to errors/changes.<sup>28</sup>

According to the March of Dimes, 1 in 5 women in the United States has her first child after age 35 and a woman's risk of having a baby with certain birth defects involving chromosomes (the structures in cells that contain genes) increases with age. For example, a woman's risk of having a baby with Down syndrome is 1 in 1,000 at age 30 and this increases to 1 in 400 at age 35 and then to 1 in 100 at age 40.

The American College of Obstetricians and Gynecologists (ACOG) recommends that all pregnant women, regardless of age, be offered a screening test for Down syndrome and certain other chromosomal birth defects (6). Screening tests are blood tests done in the first or second trimester, sometimes with a special [ultrasound](#). They help evaluate a woman's risk for having a baby with certain birth defects, but they cannot diagnose a birth defect. ACOG also recommends that all pregnant women have the option of choosing a diagnostic test, such as [amniocentesis](#) or [chorionic villus sampling](#) (CVS), instead of a screening test. The March of Dimes information also reports complications that are more common in women over 35

include: gestational diabetes, high blood pressure: placental problems, premature birth and stillbirth.<sup>29</sup>

### **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.

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 (2009) contract awards totaling less than \$900,000. This funds 6 fee-for-service providers and two population-based programs, impacting capacity, access, and the availability of educational information and outreach.

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.

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. 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. The NCHPEG also recognizes the need for continuing medical education among geneticists.

## **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. The council recommends that representation include the Department of Assistive and Rehabilitative Services (DARS), specifically from the Early Childhood Intervention Services (ECI), and Medicaid. The council also recommends adding an additional Medicaid Policy Representative that understands Medicaid funding.
2. **Access** – increase access to genetic services in rural and underserved areas of the state.
  - a. Allocate funds to increase utilization of 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.
3. **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 and implement best-practices genetic education models, as funding allows.
  - b. Expand genetics CME, CNE and CEU offerings through additional educational efforts
  - c. Continue to expand professional growth and development for genetic service providers through the DSHS Texas Health Steps Provider Education modules.
  - d. 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.
  - e. Utilize teleconferencing as an education medium.
  - f. Work with schools of public health, medicine, and nursing to promote the integration of genetics into professional study and practice.
  - g. 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.
4. **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.
5. **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.
6. **Safety Net Programs** – increase funding allocation to address contract ceiling issues and to assist with plans for comprehensive education and training.
7. **Policy** – increase efforts to assure adequate and timely reimbursement for genetic services.

## End Notes

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- <sup>1</sup> [www.dshs.state.tx.us](http://www.dshs.state.tx.us) "2004 and 2008 Natality Reports, accessed on March 14, 2012
- <sup>2</sup> March of Dimes, [http://www.marchofdimes.com/baby/birthdefects\\_chromosomal.html](http://www.marchofdimes.com/baby/birthdefects_chromosomal.html), accessed on March 13, 2012
- <sup>3</sup> National Vital Statistics Reports, Vol. 60, No. 4, Table 8, page 58
- <sup>4</sup> Centers for Disease Control, National Center for Health Statistics (CDC/NCHS), *Birth Defects/Congenital Anomalies, Cause of infant death rank, 2005*, accessed from [www.cdc.gov/nchs/data/nvsr57/nvsr75\\_14.pdf](http://www.cdc.gov/nchs/data/nvsr57/nvsr75_14.pdf) on April 9, 2010
- <sup>5</sup> [http://http://www.dshs.state.tx.us/birthdefects/data/DB\\_Data\\_report-of-Birth-Defects-Among-2000---2009-Deliveries/](http://http://www.dshs.state.tx.us/birthdefects/data/DB_Data_report-of-Birth-Defects-Among-2000---2009-Deliveries/) accessed on March 13, 2012
- <sup>6</sup> About Birth Defects in Texas, <http://www.dshs.state.tx.us/birthdefects/about.shtm>
- <sup>7</sup> Department of State Health Services, Newborn Screening Program Access Data base, report generated on May 31<sup>st</sup>, 2012. Note, figures may change from previous or future generated reports due to updates in diagnosis received from medical specialists. The 2009 birth data is from DSHS Center for Health Statistics, Vital Statistics accessed on May 31, 2012 <http://www.dshs.state.tx.us/chs/vstat/vs09/t01.shtm>
- <sup>8</sup> (ditto)
- <sup>9</sup> Department of State Health Services, TEHDI Data Reporting System to TEHDI Management Information System. accessed on 03/07/2012
- <sup>10</sup> DSHS, Texas Birth Defects Registry, *Report of Birth Defects Among 2000-2009 Deliveries*, Table 1, accessed from [http://www.dshs.state.tx.us/birthdefects/data/BD\\_Data\\_00-09/Report-of-Birth-Defects-Among-2000---2009-Deliveries/](http://www.dshs.state.tx.us/birthdefects/data/BD_Data_00-09/Report-of-Birth-Defects-Among-2000---2009-Deliveries/) on March 8, 2012
- <sup>11</sup> <http://www.dars.state.tx.us/ecis/eligibility.shtm> accessed on March 23, 2012
- <sup>12</sup> Department of Assistive and Rehabilitative Services (DARS), *DARS Autism Program*, accessed from <http://www.dars.state.tx.us/stakeholders/autism/index.shtml> accessed on March 26, 2012
- <sup>13</sup> DSHS, *Children WITH Special Health Care Needs Services Program*, accessed from <http://www.dshs.state.tx.us/cshcn/default.shtm>, on March 6, 2012
- <sup>14</sup> Department of Aging and Disability Services (DADS), *Overview of DADS Long-Term Services and Supports*, accessed from agency staff, on May 31, 2012

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<sup>15</sup> Data from Texas Medicaid and CHIP in Perspective, Eighth Edition, January 2011, HHSC, pages 1-2 and 1-3, as well as page titled "The Medicaid Numbers" for the total dollars spent in Medicaid in FFY 2009

<sup>16</sup> Medicaid and CHIP information provided by Medicaid Division, Policy Development, Health and Human Services Commission, reviewed and received on April 17, 2012

<sup>17</sup> Newborn Screening Support Group, Newborn Screening Benefits Program Data Base, accessed on April 24, 2012

<sup>18</sup> U.S. Census Bureau, *Health insurance Coverage Status and Type of Coverage by State and Age for All People: 2010*, accessed from <http://www.census.gov/hhes/www/cpstables/032011/health/toc.htm>, on April 12, 2012

<sup>19</sup> Employees Retirement Income and Security Act of 1974. (ERISA)

<sup>20</sup> U.S. Census Bureau, *Number and Percentage of People Without Health Insurance Coverage by State Using 2 and 3-Year Averages: 2007-2008 and 2009-2010*, accessed from <http://www.census.gov/hhes/www/hlthins/data/incpovhlth/2010/tables.html>, on April, 12, 2012

<sup>21</sup> U.S. Census Bureau, *Health Insurance Coverage Status and Type of Coverage by State and Age for All People: 2010*, accessed from <http://www.census.gov/hhes/www/cpstables/032011/health/toc.htm>, on April 12, 2012

<sup>22</sup> U.S. Census Bureau, *Number and Percentage of People Without Health Insurance Coverage by State Using 2-and 3-Year Averages: 2007-2008 and 2009-2010*, accessed from <http://www.census.gov/hhes/www/hlthins/data/incpovhlth/2010/tables.html>, on April 12, 2012

<sup>23</sup> <http://www.texasalmanac.com/topics/environment/environment> accessed on May 3, 2012

<sup>24</sup> <http://txsdc.utsa.edu/Reports/2002/TexasChallenge/TxChall2002Summary.pdf> accessed on May 16, 2012

<sup>25</sup> Office of the State Demographer; <http://osd.state.tx.us/> accessed on March 20, 2012

<sup>26</sup> U.S. Census Bureau, *Number and Percentage of People Without Health Insurance Coverage by State Using 2-and 3-Year Averages: 2007-2008 and 2009-2010*, accessed from <http://www.census.gov/hhes/www/hlthins/data/incpovhlth/2010/tables.html>, on April 12, 2012.

<sup>27</sup> Source: U.S Census Bureau, Current population Survey, 2010 Annual Social and Economic Supplement, POV 46: Poverty Status by State: 2009

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<sup>28</sup> Center for Health Statistics, [www.dshs.state.tx.us/chs](http://www.dshs.state.tx.us/chs), accessed on March 7, 2012

<sup>29</sup> [http://www.marchofdimes.com/pregnancy/trying\\_after35.html](http://www.marchofdimes.com/pregnancy/trying_after35.html) accessed on May 16, 2012