"The Oklahoma Genetics Advisory Council (OGAC) endorses the *State Genetics Plan's* goals, objectives, and action steps, and recommends that resources be allotted or obtained to begin implementation."

OGAC Council Members	Affiliation
Tom Allen, DO	Oklahoma Osteopathic Association
Michael Anderson, PhD	Clergy
Edward N. Brandt, Jr., MD, PhD	Oklahoma State Medical Association & University of Oklahoma College of Public Health
Robert Brown, MD	Developmental Disabilities Services, Department of Human Services (DHS)
Nancy Carpenter, PhD, Chair	HA Chapman Institute of Medical Genetics
James Coldwell, MD	Metabolic Specialist
Patti Davis	Oklahoma Hospital Association
David Domek, MD	Integris
Kevin Donnovan, MD	Ethicist, University of Oklahoma Health Sciences Center (OUHSC)
Darlene Dunn	March of Dimes
Opal Ellis	Insurance Commissioner Carroll Fisher's Representative
Maralee Hamm	Consumer
Susan Hassed, MS	OUHSC
Elisa Lee, PhD	University of Oklahoma College of Public Health
James Lewis, MD	Indian Health Service
Honorable Al Lindley	Oklahoma House of Representatives
Lynn Mitchell, MD	Oklahoma Health Care Authority
Billur Moghaddam, MD	HA Chapman Institute of Medical Genetics
John J. Mulvihill, MD, Vice-chair	American Academy of Pediatrics, OUHSC
Barbara Neas, PhD	University of Oklahoma College of Public Health
Joan Parkhurst, MD	Pediatric Hematologist/Oncologist, OUHSC
Everett R. Rhoades, MD	Native American Prevention Research Center University of Oklahoma College of Public Health
Burhan Say, MD	HA Chapman Institute of Medical Genetics
Dewey Scheid, MD	Family Practice Provider, OUHSC
John Stanley, MD	Maternal Fetal Medicine Provider, OUHSC
Kathy Stephens	Consumer
Linda A. Terrell, MHR	Consumer
Joan Walker, MD	American College of Obstetrics and Gynecology, OUHSC

Board Certified in Genetics

Oklahoma State Genetics Plan

Executive Summary

The *State Genetics Plan* was a collaborative effort between the Oklahoma State Department of Health (OSDH) and the Oklahoma Genetics Advisory Council (OGAC) and its committees. Funding for the project was provided by a federal grant from the Maternal Child Health Bureau, Health Resources and Services Administration (HRSA). Numerous public health documents and Web sites were reviewed to assist in the development of a comprehensive plan including the U.S. Department of Health and Human Services *Healthy People 2010* initiative, the Maternal and Child Health Bureau *All Aboard the 2010 Express: a 10 Year Action Plan for Children with Special Health Care Needs and Their Families,* Centers for Disease Control (CDC) Genetics and Disease Prevention, Council of Regional Networks for Genetic Services (CORN), Human Genome Project, and the American Academy of Pediatrics. The 2001 statewide genetics needs assessment also provided guidance for development. Public comment was sought through posting the action plan on the Oklahoma State Department of Health Web site and comments were requested through a mailing to Family Voices subscribers and to other public health genetic programs throughout the United States. This comprehensive document provides policymakers and genetic stakeholders an overview of the history and current directions of genetic medicine, the local and national legislation on genetic discrimination, and a review of the important role public health must play in this exciting field of medicine and health.

Advances in genetics through the Human Genome Project challenge the traditional medical model. The anticipated paradigm shift from illness care to preventive medicine interfaces with public health's long history in health promotion and disease prevention. The Human Genome Project is the science that is thrusting public health into the molecular age of health maintenance. A public health genetics program soon will be as important in disease prevention as immunization programs are today. However, scientific advances come with tough ethical, legal, and social issues. Concerns about insurance and employee discrimination based on genetic testing have been documented and must be monitored and addressed to ensure Oklahomans benefit from genetic medicine without discrimination or exploitation occurring or fear that they will occur. The establishment of a sustainable public health infrastructure to plan, implement, monitor, and evaluate genetics will be key to ensuring Oklahoman's benefit from the advances emanating from the Human Genome Project. The *State Genetics Plan* is a five-year action plan with the following mission and goals:

Mission

Effective public health strategies will ensure access to quality and timely genetic information, screening, education, and family-centered comprehensive services.

Goal 1 Educate providers, policymakers, insurance providers, medical/health career students, the public, affected families, and university and high school students regarding genomics, local genetic resources, genetic services (availability, access, indications, and benefit) and the process of referring for genetic services.

Goal 2 Develop and maintain a responsive public health genetics program to plan, implement, monitor, and evaluate genetics education and services in Oklahoma.

Goal 3 Maximize the quality of genetic testing and the effectiveness of public health screening programs to serve all the citizens of Oklahoma.

Implementation of the *State Genetics Plan* will enhance the capacity of the public health genetics programs to meet the needs of Oklahomans. For more information or comments, please contact the State Genetics Coordinator at (405) 271-6617. For information about local genetic clinical and public health program services and resources, visit the OSDH genetics Web site at http://www.health.state.ok.us/program/gp.

Who is Affected by Genetics?

Genetics is a rapidly expanding field that influences the practice of medicine throughout the lifecycle from before conception through adulthood. This field addresses hereditary disorders, birth defects, and complex diseases where both genetic and non-genetic factors play a role in the development of a disorder. Although the single gene disorders are rare, collectively they comprise over 15,500 recognized genetic disorders and affect 13 million Americans (Doyle 1).[12] A single gene disorder results when a mutation causes the product of a single gene to be altered or missing such as sickle cell disease or phenylketonuria (PKU). Although this figure is significant, the broader view of "genomic" medicine will provide opportunities for health promotion and preventive medicine throughout the lifecycle. Genomics is a new term whose standard definition is evolving; however, the Centers for Disease Control and Prevention (CDC) use the term genomics to refer to "new information emanating from the Human Genome Project" (*FAQ's* 2).[18] Advances from the Human Genome Project promise to revolutionize the practice of medicine. Just as immunizations (from polio to influenza vaccinations) are universally accepted as effective preventive medicine throughout the lifecycle, genetics will soon be considered an essential component of preventive medicine for all Americans. Dr. Victor McKusick reports genomic medicine will render medicine "more predictive and, therefore more preventive" (2294).[33] Comprehensive "genome screens" for recognition of an individual's susceptibility to common disorders can be foreseen. Clinical diagnosis will become more specific and precise, and treatments more specific and safer (McKusick 2294).[34] Genomics will provide opportunities for the individualization of medical care to achieve the "right treatment for the right patient" (McKusick 2294).[35] Better understanding of an individual's genomic make-up should permit drug therapy to get away from the one-size-fits-all approach. It should allow the selection of drugs to be more effective in the treatment of a given disorder in a given individual; thus improving treatment outcomes and decreasing the risk for adverse side effects. The development of new genomic-based drugs are expected to produce completely new lines of medications for disorders that are currently not treatable, and provide more effective and safer alternatives for drugs that are currently in use today (McKusick 2294).[36] Genomic medicine will enhance medical care to include presymptomatic identification of susceptibility to disease, preventive interventions, selection of pharmocotherapy, and individual design of medical care based on genotype. Genomic medicine promises to improve and promote the health of all citizens.

The Stottlemyre Family meets with a geneticist for diagnosis and management of a common genetic disorder in their family.

[12] *Doyle, Debra, ed. . .*
[18] *FAQ's About Workforce. . .*
[33] *The Anatomy of Human Genome...*
[34] *The Anatomy of Human Genome...*
[35] *The Anatomy of Human Genome...*
[36] *The Anatomy of Human Genome...*

Genetics Throughout the Lifecycle

Stage of Life		Genetic Service
Preconception		A couple receives genetic counseling regarding their risk for having a child with sickle cell disease. Also discussed are ways to decrease the risk for birth defects (such as folic acid to reduce the chance of neural tube defects, and that smoking has been related to a higher chance of prematurity and low birth weight). For optimal birth outcome, every couple should have preconception counseling and genetic testing offered as indicated.
Prenatal		A pregnant women gives blood to screen for birth defects. Every woman should be offered appropriate genetic counseling and testing. Offering screening for neural tube defects and Downs syndrome or determining carrier status for genetic disorders such as sickle cell disease or cystic fibrosis is standard medical practice.
Newborn		A newborn has been identified through newborn screening with the genetic disorder phenylketonuria (PKU). PKU is difficult to detect without a blood test and if left untreated can result in profound mental retardation. Newborn screening prevents mental retardation through early identification and treatment. In Oklahoma, every newborn is required to be screened for the genetic disorders of PKU, galactosemia, sickle cell disease, and for congenital hypothyroidism.
Child		A family receives genetic counseling about their child's risk for neurofibromatosis (NF) and recommendations for medical management for affected family members. Families with NF are referred to a geneticist for diagnosis and multi-disciplinary management for optimal health outcomes.
Adult		A women receives genetic counseling regarding her risk for breast cancer and possible genetic testing. Key components of the counseling session will include: risk analysis, pros and cons of genetic testing, preventive treatment options such as prophylactic surgery or chemoprevention, and the implications of testing for the woman and her family.

United States Data on Genetics and Birth Defects Throughout the Lifecycle

Preconception/ Prenatal

- 1 in 10 women do not know folic acid can prevent birth defects.[1]
- 12% of pregnant women receive inadequate prenatal care.[1]
- 3.5% of women drank heavily during pregnancy.[2]
- 13% of women smoke during pregnancy.[3]

Infancy

- 3 to 5% of all newborns have congenital malformations.[4]
- 0.5% of all newborns have a chromosomal abnormality.[4]
- 7% of all stillborns have a chromosomal abnormality.[4]
- 20-30% of all infant deaths are due to genetic disorders.[5]
- 30-50% of post-neonatal deaths are due to congenital malformations.[6]
- Alcohol-related damage (i.e., mental retardation) in 50,000 infants could be prevented if pregnant women did not drink.[2]
- 4 million newborns are annually screened at birth for treatable genetic conditions.[7]
- 10% of infant deaths (estimated) could be prevented if pregnant women did not smoke.[3]

Children

- 11.1% of pediatric hospital admissions are children with genetic disorders & 18.5% are children with other congenital malformations.[8]
- 50% of mental retardation has a genetic basis.[9]

Adult

- 12% of adult hospital admissions are due to genetic causes.[9]
- 15% of all cancers have an inherited predisposition.[10]
- 10% of the chronic diseases (heart, diabetes, and arthritis) that occur in adults have a significant genetic component.[11]

References

1. "Infant Health Statistics." Health Library March 2002. March of Dimes. 26 March 2002. <http://www.modimes.org/healthlibrary/>
2. "Drinking During Pregnancy." Health Library March 2002. March of Dimes. 15 August 2002. <http:www.modimes.org/healthlibrary/>
3. "Smoking During Pregnancy." Health Library March 2002. March of Dimes. 26 March 2002. <http:www.modimes.org/healthlibrary/>
4. Robinson A. and Linden MG. 1993. Clinical Genetic Handbook, Boston, Blackwell Scientific Publications.
5. Berry RJ, Buehler JW, Strauss LT, et al. 1987. Birth weight-specific infant mortality due to congenital abnormalities, 1960 and 1980. Public Health Report 102:171-81.
6. Hoekelman RA, Pless IB. 1988. Decline in mortality among young America during the 20th century: Prospects for reaching national mortality reduction goals for 1990. Pediatrics 82:582-95.
7. Sorrentino J. "Stepping up to the challenges of expanded newborn screening." 2 February 2001. <www.geneletter.com>
8. Scriver CR, Neal JL, Saginur R, and Clow A. "The frequency of genetic disease and congenital malformation among patients in a pediatric hospital." *Canadian Medical Association Journal* 108 (1973): 1111-15.
9. Emery AEH, and Rimoin DL. Principles and Practice of Medical Genetics, Second Ed. NY: Churchill Livingstone, 1990.
10. Schneider KA. Counseling about Cancer: Strategies for Genetic Counselors. Dennisport, Massachusetts: Graphic Illusions, 1994.
11. Weatherall DJ. The New Genetics and Clinical Practice, Second Ed. Oxford: Oxford University Press, 1985.

Oklahoma Data on Genetics and Birth Defects Throughout the Lifecycle

Preconception/Prenatal

- 64% of women reported that a physician talked to them before they got pregnant about how smoking could affect a pregnancy.[1]
- 64% of women reported that a physician talked to them before they got pregnant about how alcohol could affect a pregnancy.[1]
- 79% of women reported that they have heard or read that taking folic acid can prevent some birth defects.[1]
- 80% of women reported that a physician talked to them during a prenatal visit about how smoking during pregnancy could affect their baby.[1]
- 80% of women reported that a physician talked to them during a prenatal visit about how drinking alcohol during pregnancy could affect their baby.[1]
- 90% of women reported that a physician talked to them during a prenatal visit about the kinds of medicines that were safe to take during pregnancy.[1]
- 74% of women reported that a physician talked to them during a prenatal visit about how using illegal drugs could affect their baby.[1]
- 77% of women reported that a physician talked to them during a prenatal visit about doing tests to see if their baby had a birth defect or genetic disease.[1]

Infancy

- 100% of newborns (approximately 49,000) are annually screened at birth for treatable genetic conditions.[2]
- 3.7% of live births have a birth defect.[3]
- 0.2% of live births have a chromosomal abnormality.[3]
- 3.9% of stillbirths had a chromosomal abnormality.[3]
- 21.4% of infants who died at less than one year of age had a congenital anomaly.[3]

Children

- 4% of hospital discharges of children 19 years of age and younger are related to a genetic disease or birth defect.[4]

Adult

- 14.4% of hospital discharges of adults greater than 19 years of age are related to a genetic disease or birth defect.[4]

1. Oklahoma State Department of Health (1999). Oklahoma Pregnancy Risk Assessment Monitoring System (PRAMS).
2. Oklahoma State Department of Health Newborn Screening Program.
3. Oklahoma State Department of Health (1999). Oklahoma Birth Defects Registry Data from 1994 through 2000.
4. Oklahoma State Department of Health (1999). Oklahoma Hospital Discharge Data.

History of Genetics at the Oklahoma State Department of Health and the Oklahoma Genetics Advisory Council

The Oklahoma State Department of Health has a long history of serving Oklahomans through public health genetic programs. The first genetics program was the newborn screening program. This genetics program was established to prevent mental retardation by screening all Oklahoma infants at birth for the rare metabolic disorder phenylketonuria (PKU). The PKU newborn screening program began as a pilot project in 1963, and was so successful at identifying infants with PKU it became a state law in 1965. This early screening program enlarged to embrace the genetics and birth defects program (i.e., neural tube defect prevention program, preconception program, prenatal screening program), teratogen prevention (i.e., fetal alcohol syndrome program), the birth defect surveillance program, and expanded the newborn screening program to screen all newborns for congenital hypothyroidism, galactosemia, sickle cell disease, and hearing loss.

Today, the state genetics program is housed administratively within Screening and Special Services (SSS) of the Family Health Services of the Oklahoma State Department of Health and has a full-time State Genetics Coordinator. The SSS administers the various childhood screening programs: Newborn Screening Program (metabolic and hearing), the Oklahoma Birth Defects Registry, the Oklahoma Childhood Lead Poisoning Prevention Program, and the Genetics Program. This administrative structure provides close collaborative ties among important public health genetic programs. However, with the burgeoning advances in genetics through the Human Genome Project, public health genetics goes beyond the traditional maternal and child health programs. To address this exploding facet of medicine that promises to improve the health of citizens throughout the lifecycle, the Family Health Services of the Oklahoma State Department of Health assisted in establishing a diverse advisory group for the Commissioner of Health, the Oklahoma Genetics Advisory Council (OGAC). In October 1999, OGAC was established with 44 council and ex-officio members that included representatives from genetic providers, oncologists, family practice, state medical associations, state legislature, clergy, consumers, families with genetic disorders, the Department of Human Services Children with Special Health Care Needs program staff, and public health programs from newborn screening to chronic disease. OGAC quickly established six working committees expanding membership to 84 additional stakeholders including such diverse disciplines as mental health and a high school biology teacher. Each committee of OGAC is required to have consumer representatives; however, to strengthen the link to families affected by genetic disorders, OGAC established a Family Advisory Committee in 2002. Today, OGAC is a thriving organization that meets three times a year with a strong membership of 28 council members appointed by the Commissioner of Health and 16 ex-officio members representing diverse public health programs including the State Epidemiologist and the minority health representative. OGAC provides a model for population-based governance of genetics and has been recognized nationally for its organizational structure (Mulvihill et al. n. pag.).[39] The following active committees meet several times each year, and at each OGAC meeting, committees report, make recommendations, or seek guidance from the experts on OGAC:

- Newborn Screening Programs and Pediatrics
- Genetics Education Committee of Oklahoma (GECO)
- Adult
- Family Advisory
- Public Health Policy
- Evaluation
- Birth Defects Registry, Prenatal Screening and Diagnosis

[39] *A model for population-based. . .*

To facilitate the development of this *State Genetics Plan*, OGAC and its committees, with a total membership of 98, were involved throughout the needs assessment and development process.

The *State Genetics Plan* is a five-year plan that provides a map to ensure Oklahomans will benefit from the clinical advances in genetics. As outlined in the plan, the action steps call for an active public health role in education, data integration of public health children's health information systems, development of a comprehensive follow-up program for the newborn screening program (metabolic and hearing), community networking and partnering to ensure medical homes are accessible to serve children with special health care needs, and infrastructure development to ensure Oklahomans have access to quality genetic services in an environment free from discrimination or privacy breaches. Through continued community partnering and guidance from the Oklahoma Genetics Advisory Council, the OSDH Genetics Program will strive to meet the needs of the community through the development of effective public health strategies to assure access to quality and timely genetics information and services.

From Peas to the Human Genome Project

Medical genetics is a relatively new field of science and medicine and even newer to public health. The cornerstone of genetic science can be traced to 1865 when Gregor Mendel, an Austrian monk, discovered the principles of heredity through his experiments with garden peas. However, his discovery was unnoticed until the beginning of the 20th Century. In the 1940s, scientists began to understand the biochemical role genes play in life processes and discovered that genes were composed of deoxyribonucleic acid (DNA). The first human disease found to have a chromosomal error, Turners Syndrome, was described clinically in 1938 by Henry H. Turner, a University of Oklahoma (OU) medical professor. By the late 1950s, techniques for the scientific study of human chromosomes had been developed, and researchers began to explore the role of chromosomes in sexual development and of chromosome abnormalities as causes of abnormal physical development and reproductive problems. In 1953, James Watson and Francis Crick described the molecular structure of DNA. These early genetic discoveries provided the foundation for the success of the Human Genome Project to sequence the human genome.

The Human Genome Project was initiated in 1990 as a collaborative project between the U.S. Department of Energy and the National Institutes of Health (NIH) with the goal to map and sequence the human genome, the genetic roadmap of mankind. The Project's technology and resources have had great influence in biomedical research and are expected to vastly transform today's biological research and clinical medicine. In the continuing search for genes for various genetic conditions, researchers have benefited enormously by the improved detail of new genome maps. Myotonic dystrophy, fragile X syndrome, neurofibromatosis types 1 and 2, inherited colon cancer, Alzheimer's disease, and familial breast cancer are all genetic conditions which are now being studied by a new and improved molecular medicine. Characterized less by treating symptoms and more by looking to the most fundamental causes of disease, molecular medicine has brought hope for: 1) improving the diagnosis of these various diseases; 2) earlier detection of any genetic abnormalities inclined to disease; 3) new classes of medicine based on a reasoned approach rather than the traditional trial-and-error method; 4) genetic tests that will indicate which medication is specific to the patient's condition, instead of acting based on an educated guess; 5) safer drugs; 6) and gene therapy (*Potential* 1).[47]

Genetic medicine will improve diagnosis and prediction of disease, assessment of disease susceptibility, and provide new treatment and prevention opportunities. From peas to the mapping of the human genome, the clinical application of genomics holds dramatic and great promise to change the practice of medicine.

[47] *Potential Benefits...*

The **Human Genome Project** is the international effort to understand the hereditary instructions that make each of us unique. Understanding the human genome is important, because genes do not only influence what we look like, but genes influence what diseases we are susceptible to or may get later in life. This project promises to revolutionize medicine through:

Genetic report cards: a risk assessment that allows individuals to understand their genetic makeup and risks and provide an opportunity for each person to be educated on their genetic risk and preventive measures to decrease those risks.

Personalized medicine: the ability to offer drugs tailored to the individual makeup of the patient to avoid toxicity.

Gene and Drug Treatments: new treatment modalities to remove or cure pathologies.

The Human Genome Project

The Human Genome Project has for all practical purposes successfully sequenced the human genome. To understand how this relates to health and disease, Dr. Alan Guttmacher provides a helpful analogy: "we are learning the genetic alphabet … Knowing the alphabet is incredibly important. But just knowing the alphabet in and of itself doesn't have much impact. The important thing is knowing how the letters go together to make words (how pieces of DNA go together to make genes). To understand how the words interact to make sentences is to figure out how the genes interact with the environment to affect health and disease."

Guttmacher, A. Proceedings of the Impact of Genetic Issues on Child Health Policy. Genetics Service Branch, Maternal Child Health Bureau, Health Resources and Services Administration. Rensselaerville, New York. 21-23 June 2000.

What are Genetic Services, an Overview

Genetics refers to many aspects of health promotion and disease prevention. It can refer to inborn variation in susceptibility to disease, and to disorders that are passed on in families. It includes complex conditions such as cancer, heart disease, and diabetes, where genetic and non-genetic factors play a role in the development of a disorder. Genetics embraces birth defects and their causes, such as chromosomal disorders, neural tube defects, and fetal alcohol syndrome. Medical genetics has four major areas of specialization: the study of chromosomes (cytogenetics), the study of the structure of DNA (molecular), the function of genes (biochemical), and the medical application to diagnosis and patient care (clinical genetics). The benefits of clinical genetics and genetic testing (molecular, biochemical and cytogenetics) are in their infancy and hold great promise for the prevention of morbidity and mortality, as well as for more targeted medical management than is now possible.

Clinical Genetics

Clinical genetics began as a small sub-specialty field of pediatrics dealing with dysmorphology, the diagnosis of children with unusual features or syndromes. With the development of chromosome studies, geneticists were often consulted about whether such expensive chromosome studies should be performed. With the technological advances and decreased costs of genetic testing and screening, the consultative role of the geneticist expanded to interpretation of results, the management of rare disorders, and the continued role as detective to determine diagnoses in complex cases. Today, the practice of clinical genetics is a family-oriented care approach that provides a range of services throughout the lifecycle aimed at health education, management, prevention, and integration with primary care and sub-specialty care. Genetics is unique in that it deals with health issues for an individual and their family. This branch of medicine is committed to providing the necessary resources to communicate effectively with families about their genetic risks, especially through the utilization of genetic counselors. A board certified or board eligible genetic counselor is a health professional with a specialized graduate degree in medical genetics and counseling. The American Board of Genetic Counseling provides certification for this profession. A genetic counselor is well trained to provide information and support to families who have members with birth defects or genetic disorders and to families who may be at risk for a variety of inherited conditions. Counseling services include identification of families at risk, investigation of problems present in the family, interpretation of information about a disorder, assessment of inheritance patterns, communication of risks, and review of available options and services (*Career* 1).[6] The genetic clinical service team approach provides essential medical, laboratory, and counseling services to diagnose and manage complex disorders.

[6] *Career Information…*

Clinical Genetic Services Identifies Lethal Genetic Condition in 12-Year-Old Boy

A 12-year-old boy came to the OU Medical Center-Children's Hospital for removal of metal plates used in repair of pectus excavatum, a depression of the chest. The surgery had been performed three years previously; however, the mother had failed to have the plate surgically removed at the appropriate time. After surgical removal of the plate, the surgeon requested a consultation with Genetics to determine if this tall thin young man had the genetic disorder Marfan syndrome. There is not a genetic blood test for the disorder, therefore a geneticist must rely on clinical experience and coordinate the multi-disciplinary diagnostic workup. The genetics team evaluated the child; however, the physical exam and family history were impeded due to surgical pain and the child's current placement in foster care. Although the family history was sparse it was reported that his mother was of above average stature and two half-brothers were also tall. The child was unable to tolerate the typical physical examination required for diagnosis of Marfan syndrome; however, the limited physical examination was suggestive of the condition. The geneticist then ordered the routine work-up to rule-out Marfan syndrome that includes consultations with ophthalmology and cardiology. Those appointments were scheduled to follow a few weeks after discharge from the hospital. However, the geneticist insisted the cardiology consult and echocardiogram be performed prior to discharge due to some respiratory symptoms and the distance the center was from their home. The cardiology consultation was performed and the echocardiogram revealed a massively dilated aorta of 6.7 cm. This could result in sudden death with exercise or activity. Repair was scheduled and surgery performed. The child had a post-surgical stroke and required physical therapy, and he is expected to fully recover. The ophthalmology evaluation was negative for signs diagnostic of Marfan syndrome. Further detailed physical examination did support the diagnosis of that condition and preventive health strategies to prevent cardiac collapse and family support information were provided. However, the story does not end there. With genetics you have to treat the whole family. The two half-brothers were examined and, although tall, were not diagnosed with Marfan syndrome.

Marfan syndrome is a heritable disorder of the connective tissue that affects many organ systems, including the skeleton, lungs, eyes, heart and blood vessels. This condition affects 1 in 5,000 men and women of any race or ethnic group. A person with Marfan syndrome may be at severe risk of sudden death unless they are diagnosed and obtain proper treatment because the aorta may dissect without warning (National Marfan 1).[42]

[42] *National Marfan Foundation...*

Providers in genetics are called geneticists who have received board certification in clinical and laboratory genetic services. Their professional degree may be a MD, DO, PhD or masters. Before obtaining board certification, genetic professionals must meet a certain criteria in relation to their position in the genetic field (Wisconsin State Department of Health 8-9):[56]

Clinical Geneticists - physicians with training in a primary specialty, such as pediatrics, obstetrics, internal medicine, and who have additional subspecialty training in the clinical aspects of genetics

Clinical Biochemical Geneticist - specialists in the diagnosis and treatment of inborn errors of metabolism

Clinical Molecular Geneticists - experts in the use of molecular (DNA) tools in the diagnosis of genetic processes

Clinical Cytogeneticists - experts in the utilization and interpretation of chromosome analyses

Ph.D. Geneticists - scientists with clinical training to handle complex scientific issues in clinical genetics

Genetic Counselors - masters-level degree with training in all aspects of medical and clinical genetics as well as in counseling

American Board of Obstetrics and Gynecology, Division of Maternal and Fetal Medicine provides certification for:

Maternal Fetal Medicine Providers - physicians with training in a primary specialty of obstetrics and additional subspecialty training in the clinical aspects of prenatal/obstetrical genetic care

In addition, genetic services may be provided by other health care providers, such as nurses with advanced training in genetics.

[56] *Genetics Services Plan for Wiscon…*

What Makes Genetic Testing Different?

Prediction: Traditional medical tests inform about the patient's present condition, while genetic tests "inform" about a possible future condition.

Information beyond the patient: Genetic tests affect other individuals who have not chosen to undergo testing.

Genetic Testing

Many genetic disorders can be identified by a blood test. The three types of genetic blood tests include molecular, biochemical, and cytogenetics. Molecular (DNA) testing is the newest and most sophisticated of genetic testing and it involves the direct examination of the DNA molecule itself and is the focus of the Human Genome Project. Molecular testing can provide information about a particular DNA sequence or the presence or absence of a particular gene. Biochemical genetic testing focuses on gene products such as enzymes and other proteins. Cytogenetic testing looks at the number and characteristics of chromosomes. Genetic tests serve as useful tools, and medical practitioners might request genetic testing for the following reasons:

- To perform pre-implantation genetic diagnosis for the selection of healthy embryos for implantation
- To confirm a diagnosis of an existing condition
- To conduct prenatal testing
- To screen newborns
- To identify carriers of genetic mutations
- To perform **presymptomatic** testing (predictive for a disorder)
- To perform **predisposition** testing (inform about a possible future condition)

Tests for adult-onset disorders like breast cancer (predisposition) and Huntington disease (presymptomatic) already are widely used, and state public health programs screen newborns for a variety of genetic diseases such as phenylketonuria (PKU) and sickle cell disease. However, these applications only hint at the potential of genetic testing technology (Johnson 1).[26] Technological advances in genetic testing will allow providers to identify patients at risk for the leading causes of death in America (e.g., cancer and heart disease) and provide information on opportunities to decrease those risks. A doctor will be able to order genetic testing to determine the most effective and least toxic drug therapy. Improving drug therapy is an important aspect of genomics research since "more than 100,000 people die each year from adverse responses to medications that are beneficial to others" (Casey 2).[7]

Genetic testing opportunities are exciting and hold great promise in health promotion. A critical component of genetic testing is genetic counseling. Genetic testing must be offered and performed in an ethical and informed environment where the implications of testing, limitations of the testing, and the results of testing are communicated accurately and in a non-directive manner so that people can make informed health decisions.

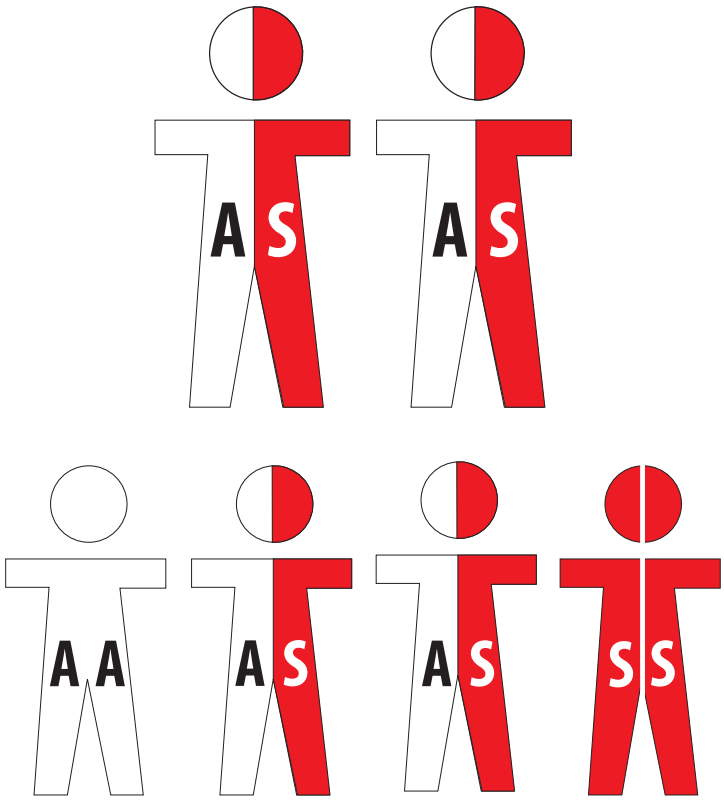
---

[26] *Johnson, Alissa...*

[7] *Casey, Denise K...*

Genetic Testing may be Useful and Necessary Throughout the Lifecycle

Preconception/prenatal: In the 1970s, screening women during the first trimester of pregnancy for neural tube birth defects by testing maternal serum for alpha feto-protein was begun, and the same technology was later shown to screen for infants with Down syndrome and other genetic disorders. Screening couples prior to pregnancy to identify carrier status for autosomal recessive genetic disorders began in the 1970s for Tay Sachs disease and sickle cell anemia. Such testing detects if a person carries a gene for a genetic disorder, but is not affected. In 2001, the American College of Obstetricians and Gynecologists (ACOG) recommended DNA screening for cystic fibrosis, an autosomal recessive disorder, to all couples seeking preconception or prenatal care. Tay Sachs disease, sickle cell anemia, and cystic fibrosis are disorders that occur in offspring of parents who are both carriers of an abnormal gene. Screening couples for carrier status allows them to make informed decisions about future reproduction. For example, if one parent is a carrier and the other is not, then their children are not at risk for a disorder. If both parents are carriers, then there is a 1 in 4 chance with each pregnancy to have a child with the disorder (autosomal recessive inheritance). Identification of disorders preconceptionally or during pregnancy through genetic testing will only increase and provide significant opportunities to prevent morbidity and mortality.



Autosomal Recessive Inheritance - Sickle Cell Anemia (Hemoglobin SS Disease)

Both parents have sickle cell trait (hemoglobin AS). They each possess one gene for normal hemoglobin (A) and one gene for sickle hemoglobin (S). Since both parents are a carrier for sickle cell trait, there is a risk for having a child with sickle cell disease (hemoglobin SS Disease). With each pregnancy there is a 25% chance that their child will have normal hemoglobin (AA), 50% chance of having sickle cell trait (AS) and 25% risk of having sickle cell anemia (SS). This risk applies to each pregnancy. There is a 1 in 4 (25%) chance of having a child with Sickle Cell Anemia (Hemoglobin SS Disease).

Newborn: Screening newborns for genetic disorders began in the 1960s, with PKU testing, and continues to be a highly successful program to prevent mental retardation. Today there are over 30 disorders for which newborns can be screened by a simple heel stick, and the future predicts the technical feasibility of screening each infant for over 1000 disorders.

Pediatric: In the newborn nursery major malformations and metabolic emergencies often warrant clinical evaluation and subsequent genetic testing. Older children may warrant genetic testing for minor malformations, subtle metabolic conditions, and growth and development delay. For adolescents, abnormal puberty and reproductive health may result in genetic testing.

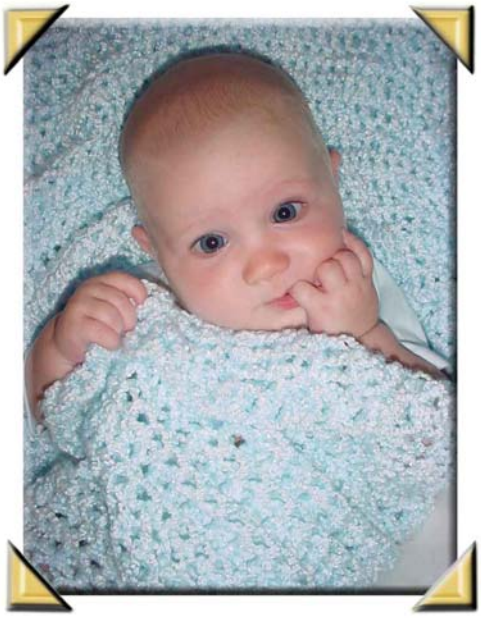
Adult: Adult presymptomatic testing for the autosomal recessive iron overload disorder, hemochromatosis, the autosomal dominant disorder of Huntington disease (HD), and tests for genetic predisposition for breast and colorectal cancers are a few examples of genetic testing in the adult. HD is often thought of when discussing adult genetics. HD is a devastating, degenerative brain disorder for which there is, at present, no effective treatment or cure. HD slowly diminishes a person's ability to walk, think, talk and reason, until he or she becomes totally dependent upon others for care, and profoundly affects the lives of entire families emotionally, socially and economically. Huntington disease is an autosomal dominant disorder with complete penetrance. This means each child of an affected person has a 50-50 chance of inheriting the gene and everyone who inherits the gene will develop the disease (*What Is* 1).[55] Huntington disease gene testing is an excellent example of the need for sensitivity and competency that must be provided when testing for genetic disorders. Implications for the individual and their family must be considered before the test is performed. Inadequate genetic counseling in the testing process for Huntington disease can result in profound consequences for the person and their family. There have been reports of healthy individuals committing suicide when they discover they have the gene for Huntington disease. These are complex counseling issues that must be considered before a genetic test is administered. Breast cancer gene testing is an example of predisposition testing. Unlike the gene testing for Huntington disease, an abnormal test result does not indicate disease or predict the inevitability that a disease will occur; it carries a risk of being affected, but does not indicate a certainty. For example, some people identified with the changed gene for breast cancer will never manifest breast cancer. Breast cancer gene testing leads to similar complex genetic counseling issues. A woman will need to consider what she will do if her test results are positive for the gene *BRCA1* or *BRCA2*, the genes linked to breast cancer. If the result is positive, what preventive measures will the woman elect to pursue? Will she elect to have a prophylactic mastectomy, or chemoprevention and increased surveillance of the breast? Will she share the test results with other at-risk family members?

[55] *What Is Huntington's...*

Genetic testing is different than traditional testing and therefore should always include a before and after genetic counseling session. During a conference meeting for the Secretary's Advisory Committee on Genetic Testing in May 2002, Dr. David Mallott noted that a patient's traditional experience with medical testing is that testing will reveal "truths" about a health condition. This perception assumes testing will provide a definite yes or no answer about a possible disorder or condition. A genetic test rarely provides yes or no answers, and typically a test result is communicated in risk. Board certified or board eligible genetic counselors have been trained to communicate such risk effectively in a non-directive manner. In addition, the person undergoing testing should understand the implications to their family. Genetic testing goes beyond the individual being tested and consideration of how that information will or will not be communicated to other family members is an important component of the counseling session. National standards on who should provide genetic counseling services have yet to be determined, but competent genetic counseling services are an important component. Studies indicate that genetic counselors are essential in the provision of comprehensive genetic services. One study reported that high-risk obstetrical patients receiving evaluation and counseling from a genetic counselor improved the detection of identifiable genetic risk factors (Koscica et al. 1033).[32] Traditionally, and as this study reports, certified genetic counselors are best prepared to communicate risk. However, there are only 2,000 genetic counselors in the U.S., and just two (2) board certified genetic counselors are currently practicing in Oklahoma. It is important that standards for genetic counseling are developed, so that Oklahomans who receive genetic services receive appropriate non-directive counseling.

Genetic Clinical and Laboratory Services in Oklahoma

Oklahoma genetic clinical and laboratory services are primarily provided in Oklahoma City and Tulsa. The two genetic centers that provide clinical, laboratory (biochemical, molecular, and cytogenetic), and genetic counseling services are the OU Medical Center in Oklahoma City and HA Chapman Institute of Medical Genetics in Tulsa and Oklahoma City branch. As of September 16, 2002, Oklahoma has five M.D. board certified geneticists who provide clinical genetic services and four genetic counselors (two board certified). Prenatal genetic services are usually provided by maternal fetal medicine providers, board certified by the American Board of Obstetrics and Gynecology, Division of Maternal Fetal Medicine. Oklahoma has nine maternal fetal medicine physicians located in Oklahoma City and Tulsa.



[32] *Assessing Genetic Risk…*

Genetic Clinical and Laboratory Services in Oklahoma
September 2002

HA Chapman Institute of Medical Genetics, Tulsa (918) 660-3838
University of Oklahoma Health Sciences Center (OUHSC), Oklahoma City (405) 271-8685

Clinical Genetic Providers

Ayesha Ahmad, MD	(918) 456-7700	Tahlequah
David Domek, MD	(405) 945-4525	Oklahoma City
Billur Moghaddam, MD	(918) 660-3838	Tulsa
John J. Mulvihill, MD	(405) 271-8685	Oklahoma City
D. Cristina Sarale, MD	(405) 236-1111	Oklahoma City
Burhan Say, MD	(918) 660-3838	Tulsa
J. Rodman Seely, MD, PhD	(405) 271-6777	Oklahoma City

Maternal Fetal Medicine Genetic Providers

(each boarded by the American Board of Obstetrics and Gynecology, Division of Maternal Fetal Medicine)

James Beeson, MD	(918) 582-0721	Tulsa
Christine Blake, MD	(918) 582-0721	Tulsa
Fred Coleman, MD	(405) 271-4665	Oklahoma City
Fred Fumia, MD	(918) 582-0721	Tulsa
Mark Harman, MD	(918) 582-0721	Tulsa
Glen Haswell, MD	(918) 742-8847	Tulsa
Asad Sheikh, MD	(405) 271-4665	Oklahoma City
John Stanley, MD	(405) 271-4665	Oklahoma City
Jeffrey Stewart, MD	(918) 747-8255	Tulsa

Genetic Laboratories

Nancy Carpenter, PhD, HA Chapman, Tulsa	(918) 660-3838
S. Terence Dunn, PhD, OUHSC, OKC	(405) 271-5249
J. Rodman Seely, MD, HA Chapman, OKC	(405) 271-6777
Shibo Li, MD, OUHSC, OKC	(405) 271-3589

Genetic Counselors

Suzanne Davidson, MS, Myriad	(918) 491-0967
Mary Floyd, CRMC, HA Chapman, Tulsa	(918) 660-3838
Susan Hassed, MS, OUHSC, OKC	(405) 271-8685
Patrick Wilson, MS, OUHSC, OKC	(405) 271-8685

Maternal Fetal Medicine Genetic Nurses, OU Medical Center, OKC

Names: Sharon Vaz, R.N. and Barbara Koop, R.N.
Phone: (405) 271-4665 or (405) 271-5026 or 1-800-937-5543

Teratogen Service 1-866-OKGENES

Hollie Hire, M.P.H., OU Medical Center, OKC (405) 271-8685

Board Certified in Genetics (directory of geneticist, by certification, in the United States can be found online at www/faseb.org/genetics)

Genetic Confidentiality And Discrimination, A Cause For Concern

The mapping and sequencing of the Human Genome has created complex ethical, legal, and social issues. While this wealth of new information has potential for great benefit, there is also potential for great harm. History has demonstrated the misuse of genetic information. The misuse of genetic information can be seen in the U.S. eugenics policies of the early 1900s that lead to discrimination, invasion of privacy, and unethical practices, such as involuntary sterilization of those determined to be "feebleminded." To avoid the exploitation and misuse of genetics that occurred at the beginning of the 20th century, the Human Genome Project established the Ethical, Legal and Social Implications (ELSI) project to anticipate and address the implications for individuals and society of the mapping and sequencing of the human genome. The ELSI project, along with numerous national and state-sponsored task forces, has been responsible for developing legislative proposals to protect genetic information and prevent discrimination. However, these efforts are slower than the scientific advances that are occurring at a phenomenal pace. As science continues to move faster than policy, court intervention is predicted to address issues of privacy infringement and discrimination. A careful review of the Oklahoma genetic needs assessment findings, national advisory group statements, current federal legislation, the executive orders from the President of the United States, and local legislation underscores the concerns that both the public and policymakers have in regard to prohibiting genetic discrimination by employers and insurance providers, and other complex issues related to genetics.

During our genetic needs assessment in Oklahoma, genetic providers were questioned regarding issues of discrimination and confidentially. It was generally reported that patients are fearful of genetic testing and often request the results be withheld from insurance providers. There were no reports of insurance or employee discrimination, but all providers agreed that their patients were anxious about these issues.

The Secretary's Advisory Committee on Genetic Testing (SACGT) was established in 1998 to advise the Department of Health and Human Services on the medical, scientific, ethical, legal, and social issues raised by the development and use of genetic tests. In a SACGT report to the Assistant Secretary of Health and Surgeon General, there was the following recommendation:

> Federal legislation is needed to prohibit discrimination in employment and health insurance based on genetic information. Federal legislation is also needed to protect the privacy of genetic information as well as other medical information in medical records. Without these protections, the public will be reluctant to undergo genetic tests that might be beneficial to its health and well-being (Enhancing 14).[14]

The Human Genome Project's Ethical, Legal and Social Implications (ELSI) project monitors legislation at the federal and state level and reports:

> No federal legislation has been passed relating to genetic discrimination in individual insurance coverage or to genetic discrimination in the workplace. Several bills were introduced during the last decade. Some of these bills attempted to amend existing civil rights and labor laws, while others stood alone. The primary public concerns are that (1) insurers will use genetic information to deny, limit, or cancel insurance policies, or (2) employers will use genetic information against existing workers or to screen potential employees. Because DNA samples can be held indefinitely, there is the added threat that samples will be used for purposes other than those for which they were gathered (Genetics Privacy 1).[19]

---

[14] *Enhancing the Oversight...*

[19] *Genetics Privacy Legislation...*

Although no specific federal genetic nondiscrimination legislation has been enacted, there are current anti-discrimination laws that may be applied to genetics including the Americans with Disabilities Act of 1990 (ADA), the Health Insurance Portability and Accountability Act of 1996 (HIPAA), and Title VII of the Civil Rights Act of 1964. The Americans with Disabilities Act prohibition of discrimination based on disability is the most likely current source of protection against genetic discrimination in the workplace. HIPPA is the only federal law that directly addresses the issue of genetic discrimination. Currently the Department of Health and Human Services are proposing modifications to the consent requirements due to the possible interference with the efficient delivery of health care. Lastly, Title VII of the Civil Rights Act might provide protection against genetic discrimination arguing racially or ethnically linked genetic disorders constitutes unlawful race or ethnicity discrimination (*Genetics Privacy* 4).[20]

On February 8, 2000, President Clinton signed an executive order prohibiting every federal department and agency from using genetic information in any hiring or promotion action. A summary of the executive order includes: 1) prohibits federal employers from requiring or requesting genetic tests as a condition of being hired or receiving benefits; 2) prohibits federal employers from requiring employees to undergo genetic tests in order to evaluate an employee's ability to perform his or her job; 3) prohibits federal employers from using genetic information to deprive them of advancement opportunities or overseas posts because of a genetic predisposition for certain illnesses; 4) and provides strong privacy protections to any genetic information used for medical treatment and research (*Genetics Privacy* 1).[21]

In 1996, the Oklahoma Legislature established a task force (OK HCR #113) to review Oklahoma House Bill No. 2478 that proposed the creation of a Genetic Nondiscrimination Act, and required the task force to report to the legislature by January 1, 1997. In 1997, this task force was extended (OK HCR #1012) for the continued review of Oklahoma House Bill No. 2478, and required the task force to report to the legislature by January 1, 1998. In 1998, HB 3169 was enacted for the purposes of preventing genetic discrimination in the workplace, prohibiting insurers from requiring or conditioning the provision of a policy by requiring or requesting an individual undergo genetic testing, and provides definitions of genetic information and genetic test. In 1999, HB 1368 was enacted for the purposes of protecting the confidentiality of individuals participating in research studies by prohibiting the subpoena or discovery of research records in civil suits, provides that stored tissues can be used for genetic research if informed consent has been obtained, and protects the confidentiality of individuals participating in research studies by prohibiting the publishing of a participants identification unless informed consent has been obtained.

The U.S. Congress has yet to pass comprehensive legislation addressing privacy of genetic information and health insurance discrimination. The federal laws of ADA, HIPPA, Civil Rights Act, and the Oklahoma state statutes are inadequate to protect genetic privacy and prevent discrimination. The provision of leadership and community partnerships to ensure all Oklahomans benefit from genetic medicine of today and of the future, and to monitor the state for genetic discrimination, invasion of privacy, or exploitation will be a crucial role for public health.

---

[20] *Genetics Privacy Legislation...*

[21] *Genetics Privacy Legislation...*

Existing Federal Anti-discrimination Laws and How they Apply to Genetics

Americans with Disabilities Act (ADA)

- Prohibits discrimination against a person who is regarded as having a disability.
- Protects individuals with symptomatic genetic disabilities the same as individuals with other disabilities.
- Does not protect against discrimination based on unexpressed genetic conditions.
- Does not protect potential workers from requirements or requests to provide genetic information to their employers after a conditional offer of employment has been extended but before they begin work (Note: this is a heightened concern because genetic samples can be stored).
- Does not protect workers from requirements to provide medical information that is job related and consistent with business necessity.

In March 1995, the EEOC issued an interpretation of the ADA. The guidance, however, is limited in scope and legal effect. It is policy guidance that does not have the same legal binding effect on a court as a statute or regulation and has not been tested in court. According to the interpretation:

- Entities that discriminate on the basis of genetic predisposition are regarding the individuals as having impairments, and such individuals are covered by the ADA.
- Unaffected carriers of recessive and X-linked disorders, individuals with late-onset genetic disorders who may be identified through genetic testing or family history as being at high risk of developing the disease are not covered by the ADA.

Health Insurance Portability and Accountability Act

- Prohibits group health plans from using any health status-related factor, including genetic information, as a basis for denying or limiting eligibility for coverage or for charging an individual more for coverage.
- Limits exclusions for preexisting conditions in group health plans to 12 months and prohibits such exclusions if the individual has been covered previously for that condition for 12 months or more.
- States explicitly that genetic information in the absence of a current diagnosis of illness shall not be considered a preexisting condition.
- Does not prohibit employers from refusing to offer health coverage as part of their benefits packages.

Limitations: Applies to employer-based and commercially issued group health insurance only. There is no similar law applying to private individuals seeking health insurance in the individual market.

HIPPA National Standards to Protect Patients' Personal Medical Records, December 2000

This new regulation will protect medical records and other personal health information maintained by health care providers, hospitals, health plans and health insurers, and health care clearinghouses. Congress mandated the regulation when it failed to pass comprehensive privacy legislation (as required by HIPPA) by 1999. The new standards: limit the non-consensual use and release of private health information; give patients new rights to access their medical records and to know who else has accessed them; restrict most disclosure of health information to the minimum needed for the intended purpose; establish new criminal and civil sanctions for improper use or disclosure; and establish new requirements for access to records by researchers and others. They are not specific to genetics; rather they are sweeping regulations governing all personal health information. In 2002, Department of Health and Human Services are proposing modifications to the consent requirements due to the possible interference with the efficient delivery of health care.

Title VII of the Civil Rights Act

- Protection is available only where an employer engages in discrimination based on a genetic trait that is substantially related to a particular race or ethnic group.
- A strong relationship between race and national origin has been established for only a few diseases.

(*Genetics Privacy* 3-5)[22]

Oklahoma HB 3169

Prohibits health insurers, for the purpose of determining eligibility, establishing premiums, limiting coverage, renewing coverage, terminating coverage or any other underwriting decision in connection with the offer, sale or renewal or continuation of a policy, except to the extent and in the same fashion as an insurer limits coverage, or increases premiums for loss caused or contributed to by other medical conditions presenting an increased degree of risk:

1. requiring or requesting any individual to obtain a genetic test; and
2. conditioning the provision of the policy upon a requirement that an individual take a genetic test.

For purposes of distinguishing between or discriminating against or restricting any right or benefit otherwise due or available to an employee or prospective employee, other than in connection with the determination of insurance coverage or benefits, no employer shall:

1. seek to obtain, or use a genetic test or genetic information of the employee or the prospective employee; or

2. require a genetic test of or require genetic information from the employee or prospective employee.

Definition of **genetic information:** Information derived from the results of a genetic test. Genetic information shall not include family history, the results of a routine physical examination or test, the results of a chemical, blood or urine analysis, the results of a test to determine drug use, the results of a test for the presence of the human immunodeficiency virus, or the results of any other test commonly accepted in clinical practice at the time it is ordered by the insurer.

Definition of a **genetic test:** A laboratory test of the DNA, RNA, or chromosomes of an individual for the purpose of identifying the presence or absence of inherited alterations in the DNA, RNA, or chromosomes that cause a predisposition for a clinically recognized disease or disorder. Genetic test does not include: (A) a routine physical examination or a routine test performed as a part of a physical examination; (B) a chemical, blood, or urine analysis; (C) a test to determine drug use; (D) a test for the presence of the human immunodeficiency virus; or (E) any other test commonly accepted in clinical practice at the time it is ordered by the insurer.

Oklahoma House Bill 1368

Provides that all research records of individual subjects in genetic research studies shall be confidential and not subject to subpoena or discovery in civil suits, except where the information in the records is the basis of the suit.

Provides that the confidentiality provisions of the bill shall not apply to an insurer or to an individual or third party dealing with an insurer in the ordinary course of underwriting, conducting or administering the business of life, disability income or long-term care insurance.

Provides that stored tissues can be used for genetic research studies if informed consent has been obtained.

Provides for the publishing or use of results of genetic research studies for research or educational purposes if no individual subject is identified or if specific informed consent from the individual has been obtained.

Definition of **Genetic Research Studies:** Those genetic research studies approved by an institutional review board as defined by 21 CFR, Section 50 or conducted subject to the requirements of the federal common rule at 21 CFR, Section 50 and Section 56, and 45 CFR, Section 46.

Genetics and Public Health – Why Does Oklahoma Need a State Genetics Plan?

The Agency for Health Care Policy and Research, Centers for Disease Control and Prevention (CDC), Health Resources and Services Administration (HRSA), and the National Institutes of Health (NIH) best describe the importance of a public health genetics program:

> Advances in genetics are rapidly increasing opportunities for understanding and promoting health, preventing disease, and lowering mortality and morbidity. It is anticipated that the growing array of laboratory procedures will allow us to screen for a variety of conditions with a genetic component and, in many cases promote health and well-being. New genetic information will be utilized to develop innovative therapeutic measures. In turn, new genetic knowledge will provide unprecedented opportunities for individuals to learn about their genetic make-up and discuss this information with their health care providers in the context of health promotion and disease prevention. Integration of genetic knowledge and technology into health policy, research, and practice represents enormous challenges and opportunities for public health leadership. New policy constructs are needed to assure the safety and effectiveness of genetic tests and their appropriate use in clinical and public health practice. Health services research is required to evaluate the clinical utility of genetic testing in populations to develop evidence for recommendations and standards of practice. Additional policies are needed to protect the confidentiality of genetic information and to prevent it from being used to discriminate or stigmatize. There is also a need to educate health professionals and policy makers about genetic technologies and information and to enhance public understanding of the benefits, risks, and limitations of genetic testing, and the meaning and implications of genetic information (Agency 1).[1]

The Genomics and Disease Prevention book (a collaborative effort between the CDC, National Human Genome Research Institute, and University of Washington) expresses a need for public health leadership:

> How to use knowledge from genetics research to promote health and prevent disease - the fundamental mission of public health - is now being explored. However, population-based information is lacking about the distribution of genotypes in different populations, the benefits and risks of genetic testing, and the efficacy of early interventions. Moreover, the complex issues that have emerged (e.g., rapid commercialization of genetic tests, quality of laboratory testing, availability of and access to interventions, and potential discrimination against and stigmatization of individuals and groups) call for public health leadership (Khoury 2).[30]

National leaders recognize the impact that genetics will have for the promotion of health and the potential problems of discrimination. Through public health core functions of assessment, policy development and assurance, state public health agencies must ensure genetic services are integrated into the health care system, incorporated into public health practice, and are accessible and utilized to benefit the health outcomes of citizens. The *State Genetics Plan* will provide a guide to ensure Oklahomans benefit from genetic advances through the establishment of a comprehensive public health program to assure that an infrastructure exists to plan, implement, monitor and evaluate genetics in Oklahoma.

The *State Genetics Plan* is organized with three goals that address the issues of (1) genetic education for health care providers and the public, (2) development of a sustainable public health genetics infrastructure to assess, implement, monitor, and evaluate genetics in Oklahoma, and (3) screening and genetic testing. Each goal has an introductory segment reviewing the literature that supports the need for public health action. Each goal has objectives and measurable action steps that were developed and prioritized in collaboration with the Oklahoma Genetics Advisory Council (OGAC) and its committees. OGAC will provide oversight during the implementation process with specific action steps being assigned to committees for action.

---

[1] *Agency for Health Care…*

[30] *Genetic and Public Health in the 21st Century…*

Goal 1 Education

Educate providers, policymakers, insurance providers, medical/health career students, the public, affected families, and university and high school students regarding genomics, local genetic resources, genetic services (availability, access, indications, and benefit) and the process of referring for genetic services.

Advances in genetics are occurring at a pace that promises to revolutionize medicine. As penicillin changed the face of medical practice in the prevention of morbidity and mortality from infectious diseases, scientists predict genomics will change the practice of medicine to focus on prevention versus illness, improve medical interventions, and increase the human lifespan to 150 years by the middle of this century. For Oklahomans to benefit from advances in genetics, an informed public, and a competent Oklahoma workforce, and access to local genetic services must be assured. The *State Genetics Plan* outlines a comprehensive action plan for statewide genetics education at every level from high school to health care providers.

For Oklahomans to benefit from advances in genetics, an informed public, a competent Oklahoma workforce, and access to local genetic services must be assured. The *State Genetics Plan* outlines a comprehensive action plan for statewide genetics education at every level from high school to health care providers.

Since most health care providers have received training prior to the advances in the genetics seen today, continuing education will be a critical component to ensure new genomic health technologies and services are appropriately integrated into the clinical and public health settings. On May 13, 2002, Dr. Eve Slater, Assistant Secretary for Health, stressed the importance of educating U.S. health care workers during her opening remarks at the Secretary Advisory Committee on Genetic Testing educational conference:

> Innovative strategies for educating today's health professionals will be important in ensuring that professionals are equipped to properly use and interpret genetic information. Understanding the science of genetics and the medical benefits of testing are only part of ensuring the appropriate use of genetic tests. Providers must also be sensitive to the ethical, legal, and social implication of genetics, including concerns about the misuse of genetic information (Slater 4).[50]

Education is essential in preparing providers to incorporate genetics into their practice and to recognize that genetic testing is potentially harmful. The Secretary's Advisory Committee on Genetic Testing reports education as an essential step to ensure citizens benefit from advances in genetics:

> "Since genetic education and counseling are critical to the appropriate use, interpretation, and understanding of genetic test results, efforts to ensure the education of the public as well as health providers about genetics is necessary" (*Enhancing* 14).[15]

---

[50] *Genetic Testing…*

[15] *Enhancing the Oversight…*

The following segment for goal 1 will provide a brief overview of the importance of public health's role in education, national guidelines on genetic education for health care providers, and the current educational strengths of the genetics program. The action steps for Education, Goal 1, begin on page 31.

A competent health care workforce is essential for the successful integration of genetics into health care practice. "Genomics-based clinical medicine will require primary care physicians to be competent in the interpretation of gene screens and in advising appropriate health measures" (McKusick 2294).[37] As the public becomes better informed about genetic testing and risk assessment, they will seek advice from primary care providers, physician assistants, nurses, and advanced practice nurses. This will result in a greater responsibility for information, use and interpretation of genetic tests by health care providers who have often had limited training in genetics (*National Coalition* n. pag.).[41] At the 1999 Second National Conference on Genetics and Disease Prevention, representatives from the Health Resources and Services Administration and the Centers for Disease Control and Prevention (CDC) discussed the need to prepare the workforce and charged public health to develop and implement genetic educational programs to prepare the workforce to understand the effects of genetics on health and disease and utilize this information to improve health and prevent disease. A national survey of public health leaders also indicated future endeavors of public health genetic programs would largely be educational (Piper et al. n. pag.).[46] In Oklahoma, a genetic needs assessment conducted by the Oklahoma State Department of Health (OSDH) Genetics Program in June 2001 included interviews with key informants who reported education as the primary role for the OSDH genetics program. In January 2002, the Oklahoma Genetics Advisory Council (OGAC) agreed that educating health care providers, policymakers, insurance providers, educators, and the public regarding genetics is an essential step towards assuring Oklahomans have access to genetic advances in medicine. National and local experts agree that an effective genetics education program for health care providers and the public is an essential component of a public health genetics program. Preparing the health care workforce to answer patients' questions about genetic testing and disease risk will be an essential role of public health.

To assist state public health agencies to implement effective education programs, the CDC and the National Coalition for Health Professional Education in Genetics (NCHPEG) developed guidelines for educating the nation's health care providers. NCHPEG is an organization sponsored by the American Medical Association, the American Nurses Association, and the National Human Genome Research Institute. NCHPEG guidelines focus on the following three educational objectives for health care professionals: (1) appreciate limitations of his or her genetic expertise, (2) understand the social and psychological implications of genetic services, and (3) know how and when to make a referral to a genetics professional (*Core* 2).[9] The CDC document, "Genomics Workforce Competencies" are guidelines developed for the public health workforce. The NCHPEG and CDC guidelines will be utilized to develop effective education programs for health care providers. Both guidelines are available on the Web.

---

[37] *The Anatomy of Human Genome...*

[41] *NCHPEG...*

[46] *The Role of State...*

[9] *Core Competencies...*

The key focus of the Genetics Program of the Oklahoma State Department of Health (OSDH) will be to facilitate the development of a well-prepared health care workforce and improving the genetic literacy of the public. The OSDH and its community partners have already taken many steps to meet this education challenge by: (1) establishing a full-time OSDH Genetics Coordinator in November 2000, (2) establishing an advisory council for the Commissioner of Health in 1999 (Oklahoma Genetics Advisory Council), (3) providing an outreach educational program, *The New Genetic Issues for Health Professionals and the General Public,* to rural communities sponsored by the Genetics Educational Committee of Oklahoma (GECO), a committee of OGAC, (4) providing the *Genetics in Primary Care,* and the *Genetics in Your Practice* seminars sponsored by the HA Chapman Institute of Medical Genetics, (5) developing and distributing a genetics health care provider directory for genetic clinical and laboratory resources with a guide on who should be referred, (6) administering the successful educational program of the OSDH newborn screening program, (7) the OSDH folic acid educational program, (8) the Oklahoma University of Health Sciences Center (OUHSC) organized ongoing health care provider education programs, such as Center for Interdisciplinary Learning and Leadership, (9) the efforts of the OUHSC to establish a masters program for genetic counselors, and (10) the Oklahoma telecommunication infrastructure. The weakness of the current public health genetics program is the lack of personnel to coordinate a statewide education campaign.

The key focus of the Genetics Program of the Oklahoma State Department of Health (OSDH) will be to facilitate the development of a well-prepared health care workforce and improving the genetic literacy of the public.

Summary

Goal 1

With 73,096 health care providers in Oklahoma, it will be important for public health to actively engage all providers, from social work to physicians, to participate in genetic education programs that will be a benefit to their patients. Increasing awareness and knowledge of genetic conditions, genetic testing (availability, access, indications and benefits, risks, and limitations), genomics, referral, ethical, legal, and social issues, and the availability of local genetic services will be essential components of the education program. Public education on genetics will also be equally important. The *State Genetics Plan's* four objectives for goal 1 include: (1) the development of an OSDH genetic resource center, (2) establishing a statewide genetics education campaign, (3) developing a collaborative program to ensure target populations for education are reached, and (4) developing a statewide preconception/prenatal public health education program. The Genetics Education Committee of Oklahoma, a committee of OGAC, identified five action steps as priority for implementation.

Goal 1 Action Plan

"Educate providers, policymakers, insurance providers, medical/health career students, the public, affected families, and university and high school students regarding genomics, local genetic resources, genetic services (availability, access, indications, and benefit) and the process of referring for genetic services."

Objective 1 - The OSDH Genetics Program, in collaboration with OGAC, will establish and maintain a genetics education **resource center** to facilitate the education of health care providers and the public on genetic services and resources.

Action Steps (◇ symbol identifies prioritized action steps):

- ◇ Develop and publicize a genetics speakers' bureau (for use at all levels from physician to high school students (priority number 5).
- ○ Develop or identify genetics educational tools for high school students.
- ○ Develop or identify a genetics self-study guide for health care providers.
- ○ Maintain and continue to develop the OSDH genetic Web site as a resource for local genetic services, genetics education for the public and health care providers, updates on technology advances, and family support and resource information (the Family Advisory Committee will provide oversight of the Web site development of the family resource section).
- ○ Develop a practitioner manual for newborn (hearing and metabolic) screening program.
- ○ Develop educational brochures about genetics:
  - i. Benefit and indications for clinical genetic services throughout the lifecycle to assist in educating insurers and policymakers.
  - ii. Genetics as a career for high school and college students.
  - iii. Teratogen hotline information to include promotional items, such as magnets.
- ○ Develop and identify existing family resource guides that include services for children with special health care needs and genetic resources.

Objective 2 - The OSDH Genetics Program will develop an organized **education campaign** for the public and health care providers to include lectures, community forums, publications, and hospital in-services.

Action Steps (◇ symbol identifies prioritized action steps):

- ◇ Secure a full-time educator (preferably a board-certified genetic counselor) for the OSDH Screening and Special Services programs of newborn screening (hearing and metabolic), birth defects registry, and genetics (priority number 1).
- ◇ Develop and provide ongoing genetic educational programs, including genomics, for public health and health care providers utilizing the national guidelines developed by CDC and NCHPEG to include (priority number 2):
  - i. Co-sponsor genetics educational efforts with GECO, MOD, HA Chapman Institute of Medical Genetics, OUHSC, and other local organizations.
  - ii. Develop a public health sponsored annual genetics seminar collaborating with professional organizations targeting physicians, specifically family practice, internist, and pediatricians.
  - iii. Collaborate with existing distance-learning programs to develop a strategy to offer genetic education programming for providers.
  - iv. Collaborate with professional organizations to include genetic presentations or articles in their publications or seminars.
  - v. Annually by the end of the first quarter, notify high schools, universities, medical/career programs, hospitals, medical/career associations, and community organizations of genetic educational offerings.
  - vi. Provide lectures upon request.
- ○ Provide a minimum total of 12 genetic lectures and/or forums to high schools, universities, medical/career programs, the public, hospitals, and medical/career associations per year.
- ○ Develop a hospital in-service program for newborn screening and birth defects registry surveillance programs of OSDH and provide every 12 to 24 months and as needed.
- ○ Develop and distribute Screening and Special Services publications that will facilitate access to services and education efforts:
  - i. Publish an annual Screening and Special Services program newsletter featuring articles on newborn screening (metabolic and hearing), birth defects registry, lead poisoning prevention, and genetics.
  - ii. Distribute the *State Genetics Plan* to policymakers, health care providers and post on the OSDH Web site.

Objective 3 - The OSDH Genetics Program will collaborate with the public, professionals, and OGAC to **maximize genetics education opportunities** in Oklahoma.

Action Steps (◇ symbol identifies prioritized action steps):

- ◇ Contact and establish relationships with insurers to provide education on reimbursement issues related to newborn screening and genetic services, such as genetic counseling, lab utilization, and newborn screening fee (priority number 3).
- ◇ Collaborate with OUHSC faculty to encourage the incorporation of genetic courses into the curriculum of medical schools, nursing, and allied health (priority number 4).
- ○ Identify opportunities to offer genetic education offerings by networking with the following organizations and programs:
  - i. Oklahoma Turning Point Initiative
  - ii. March of Dimes
  - iii. OUHSC outreach education programs
  - iv. Family Voices
  - v. Early Intervention
  - vi. Children with Special Health Care Needs Program
- ○ Develop relationships with undergraduate programs and local public high schools to advocate for the incorporation of genetics into the curriculum and publicize genetic career options.
- ○ Collaborate with Public Health Policy Committee of OGAC to identify educational opportunities for policymakers, i.e., education offerings at the Capitol.

Objective 4 – The OSDH Genetics Program will facilitate collaboration with the Oklahoma Births Defect Registry, Women's Health Service, Chronic Disease Service, OGAC Birth Defect Registry, Prenatal Screening and Diagnosis Committee, and the March of Dimes (MOD) in the development of a **statewide program of preconception and prenatal health education** to identify populations at risk and to provide education about the prevention of birth defects and adverse birth outcomes.

Action Steps:

- ○ Develop recommendations to implement prenatal screening for cystic fibrosis.
- ○ Develop a pamphlet on prenatal screening for cystic fibrosis.
- ○ Develop a public education campaign targeting high school students and family planning program clients regarding the prevention of birth defects and prematurity related to teratogen exposures such as alcohol and smoking.
- ○ Maintain and expand the public health prevention of neural tube defects campaign.
- ○ Collaborate with the update of the "Diabetes and Pregnancy Health Expectations Guidelines," and facilitate the implementation of the guidelines statewide.
- ○ Develop and implement a preconception screening tool statewide.

This is the Spannagel family.
Their story begins on page 34.

Goal 2 Public Health Genetics Infrastructure

Develop and maintain a responsive public health genetics program to plan, implement, monitor, and evaluate genetics education and services in Oklahoma.

Through the efforts of the Human Genome Project, genetics, once an obscure program for rare genetic disorders, will soon become an integral part of our health care system and provide amazing opportunities for disease prevention and health promotion. Today, public health genetics is defined as the application of advances in genetics and molecular biotechnology to improve public health and prevent disease (Khoury, Genetics n. pag.).[28] Historically, public health's established network of essential services has successfully worked to prevent diseases, provide early screening, promote and protect health, and assure access to the health care system. This network can now facilitate the integration of genetic advances into the health care system and into public health practice. Through public health core functions of assessment, policy development, and assurance, state public health agencies must, among other activities, ensure genetic services are integrated into the health care system and are accessible and utilized to benefit the health outcomes of citizens. The *State Genetics Plan* will provide the guidance to ensure Oklahoma benefits from genetic advances through the establishment of a collaborative, comprehensive public health genetics program to assure an infrastructure exists to plan, implement, monitor, and evaluate genetics in Oklahoma. Public health must proactively develop an infrastructure capacity to ensure: (1) genetic advances are incorporated into public health practice; (2) local clinical genetic services are maintained, accessible, meet standards, and have the capacity to meet the needs of all Oklahomans; (3) citizens are knowledgeable and can advocate for genetic services to improve their health; (4) providers have the tools needed to integrate genetics into their daily practice; and (5) systems exist to monitor the ethical, legal, and social issues related to local genetic services. The *State Genetics Plan* for infrastructure was developed by reviewing information and documents from the Human Genome Project, Healthy People 2010 initiative, All Aboard the 2010 Express: *A Ten-Year Action Plan for Children with Special Health Care Needs and Their Families,* publications from the Council of Regional Networks for Genetic Services, public health data system plans to create a Child Health Information System, and the statewide genetic needs assessment findings related to reimbursement for genetic services. The following segment for goal 2 will provide a brief review of these resources and an overview of the evaluation activities for the plan. The action steps for Public Health Genetics Infrastructure, Goal 2, begin on page 43.

A public health genetics program in the future will be as important in disease prevention as immunization **programs are today.**

[28] *Genetics and Public Health...*

Human Genome Project

The Human Genome Project is the science that is thrusting public health into the molecular age of health maintenance. The Human Genome Project began in 1990 as a collaborative project between the U.S. Department of Energy and the National Institutes of Health (NIH) with the goal to map and sequence the human genome. Today, several types of genome maps have been completed, and a working draft of the entire human genome sequence was announced in June 2000 (Human Genome 1).[25] Oklahoma has played a role in this exciting project through Dr. Bruce Roe's efforts to successfully sequence chromosome number 22. For more information about local efforts, visit this Web site: www.genome.ou.edu. The ultimate goal of the Human Genome Project is to "use this information to develop new ways to treat, cure, or even prevent the thousands of diseases that afflict humankind" (Medicine 1).[38] Since virtually all disease, except some cases of trauma, have a genetic component (Collins and McKusick 540),[8] the opportunities to improve the health of Americans are staggering. The anticipated paradigm shift from illness care to preventive medicine interfaces with public health's long history in health promotion and disease prevention. It is predicted that a public health genetics program in the future will be as important in disease prevention as immunization programs are today.

Since virtually all disease (except some cases of trauma) have a genetic component (Collins and McKusick 540),[8.2] the opportunities to improve the health of Americans are staggering.

The Human Genome Project is the science that will thrust public health into the molecular age of health maintence.

Healthy People 2010 and Children with Special Health Care Needs - 2010 Express

The overarching goals of the Healthy People 2010 initiative of the US Department of Health and Human Services are to "increase quality and years of healthy life" and "eliminate health disparities" (Healthy 1).[24] Technological advances in the field of genetics hold great promise to meet this challenge and fulfill the 2010 goals and purpose of promoting health and preventing illness, disability, and premature death. To facilitate the attainment of Healthy People 2010, the public health genetics program developed a comprehensive plan to address genetics from infancy to adulthood.

Although genetics impacts the entire lifecycle, the children with special health care needs (CSHCN) population will be a target population for special monitoring by the OSDH genetics program. The *All Aboard the 2010 Express* document defines CSHCN "as those children who have or are at increased risk for a chronic physical, developmental, behavioral, or emotional condition and who also require health and related services of a type or amount beyond that required by children generally." National estimates of the number of children with special health care needs range between 15 and 20 percent of all children (U.S. Department 4).[53] Of the CSHCN population, at least 10% have chromosomal problems

---

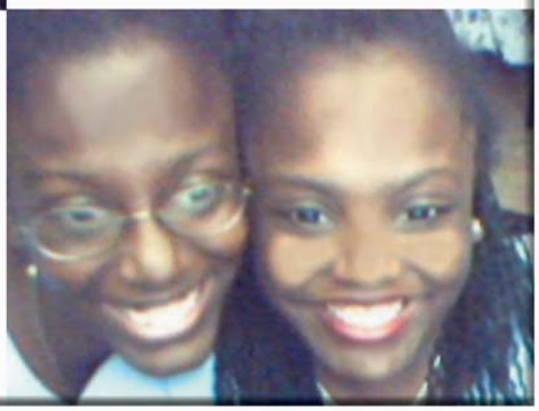
[25] *Human Genome Project…*
[38] *Medicine and the New….*
[8, 8.2] *Implications...*
[24] *Healthy People...*
[53] *US Department of Health…*

and 3% have genetic related disorders. However, considering the genetics of diabetes, asthma, cancer, and hearing loss, clearly a significant percentage of children with special health care needs have conditions with a genetic component (Cunningham, Percent n. pag.).[11] To ensure the CSHCN with genetic disorders receive needed services, the state plan outlines the development of strong links to all programs that serve CSHCN. To promote and facilitate the development of community-based, family-centered, culturally competent, and coordinated systems of care, the *2010 Express* document's six core outcomes were utilized to develop the plan's service systems for CSHCN.

The genetics program will build on the successful newborn screening program follow-up systems to enhance service systems for infants identified through public health screening programs into adulthood, with the ultimate goal to expand these service systems to all CSHCN affected by genetic disorders. The establishment of medical homes with access to needed services will be a key performance measure of the public health genetics program. A medical home is a source of routine healthcare in the community that assists in early identification, provides ongoing primary care, and coordinates with a broad range of other special, ancillary and related services. The state plan outlines a community-based approach to develop an infrastructure that operates across service sectors promoting program communication and collaboration to better meet the needs of the child and the family. The planned activities to achieve success with the *2010 Express* six core outcomes are outlined throughout the *State Genetics Plan's* action plan. Children with special health care needs in Oklahoma will benefit from the successful implementation of the plan.

The Smith sisters have a common genetic disorder prevalent in African Americans.

Kayla and Dewayna's story can be found on page 54.



[11] *Cunningham, George...*

A summary of the six core outcomes of *All Aboard the 2010 Express* and how the OSDH Genetics Program and implementation of the *State Genetics Plan* will facilitate achievement of these core outcomes:

All Aboard the 2010 Express is a 10-year action plan with six core outcome measures. The following identifies each core outcome and the related genetic program and *State Genetics Plan* activities (U.S. Department 7):[54]

1. Families of children with special health care needs will partner in decision making at all levels and will be satisfied with the services they receive.

Genetics Program: Families are currently represented on the Oklahoma Genetics Advisory Council and on each OGAC committee. A Family Advisory Committee of OGAC has been established.

2. All children with special health care needs will receive coordinated, ongoing, comprehensive care within a medical home.

State Plan: A long-term case management follow-up program will initially be established for the Newborn (metabolic and hearing) Screening Program and later expanded to include all CSHCN affected by genetic disorders.

3. All families of children with special health care needs will have adequate private and/or public insurance to pay for the services they need.

State Plan: Public health leadership and advocacy activities in collaboration with OGAC and its committees and planned case management services will assist with this outcome.

4. All children will be screened early and continuously for special health care needs.

State Plan: Public health genetic and newborn screening infrastructure development, including the maintenance of short-term follow-up activities, and planned long-term follow-up case management system and data integration projects, will provide screening and effective referral for services.

5. Community-based service systems will be organized so families can use them easily.

State Plan: Public health genetic and newborn screening infrastructure development and collaboration with OGAC and its committees will develop effective systems to serve CSHCN.

6. All youth with special health care needs will receive the services necessary to make transition to all aspects of adult life, including adult health care, work and independence.

State Plan: The planned collaborative project with the OUHSC to establish an Adult Transition Program by establishing a system to serve sickle cell disease and cystic fibrosis clients and later expanded to include all CSHCN affected by genetic disorders will facilitate attainment of this outcome.

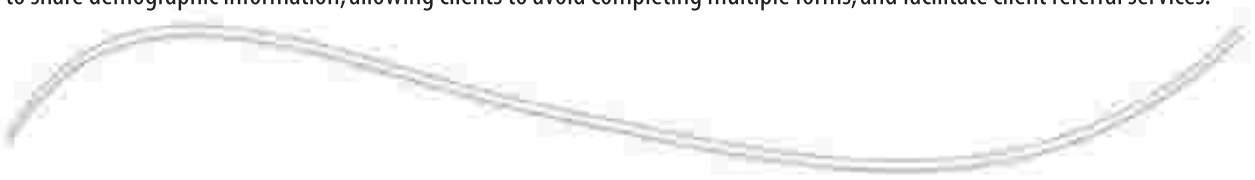
[54] *US Department of Health…*

Council of Regional Networks for Genetic Services (CORN)

In 1985, the Council of Regional Networks for Genetic Services (CORN) was formed through funding by Maternal Child Health Bureau, Health Resources and Services Administration. CORN was established to provide a forum for dialogue and national coordination among the U.S. genetic networks. In 1997, the CORN published guidelines for the development of a public health genetics program. The purpose of the guidelines was to provide state and public health agencies with an outline of suggested components for a genetic services system. CORN identified standards for genetic services and charged public health to participate in quality assurance measures to ensure all populations benefit from genetic services. Through assessment, policy development, assurance, and collaboration, state genetic programs can prevent morbidity and mortality throughout the lifecycle (CORN 14).[10] The CORN guidelines provided a useful framework in developing the action plan. CORN no longer exists today, but collaboration between states continues through the National Newborn Screening and Genetics Resource Center (NNSGRC). The NNSGRC Web site is at genes-r-us.uthscsa.edu.

Integration of Public Health Data Systems

The integration of public health data systems is an important component to the development of an infrastructure with the capacity to monitor and evaluate genetic services. The *State Genetics Plan* addresses data integration, and many projects are underway to develop an OSDH Child Health Information System (CHIS). The CHIS project has two purposes: (1) allow health care providers to access a child health profile to ascertain an infant's screening and immunization status, and (2) allow effective referral and monitoring; is the child enrolled in Early Intervention, WIC, etc? Current integration activities include the integration of three public health data systems: (1) newborn hearing, (2) newborn metabolic disorder screening program, and (3) vital records. A Newborn Hearing Screening federal grant is funding this integration project. After this project is successfully implemented, the genetics program will participate in assessing the feasibility of integrating with immunization and other public health program databases, such as WIC and Lead Screening. Additional integration efforts will involve the OSDH county health department data system PHOCIS (Public Health Oklahoma Client Information System). PHOCIS provides an overview of the services provided to each citizen by the health department. Linking with this system will provide oversight capabilities; did a child obtain Early Intervention services? Broader integration efforts will be explored and include possible linkage with the Joint Oklahoma Information System Network (JOIN) effort. JOIN is a statewide initiative to link agency information systems to avoid the duplication of data by allowing agencies to share demographic information, allowing clients to avoid completing multiple forms, and facilitate client referral services.



Public health must proactively develop an infrastructure capacity to ensure: (1) genetic advances are incorporated into public health practice; (2) local clinical genetic services are maintained, accessible, meet standards, and have the capacity to meet the needs of all Oklahomans; (3) citizens are knowledgeable and can advocate for genetic services to improve their health; (4) providers have the tools needed to integrate genetics into their daily practice; and (5) systems exist to monitor the ethical, legal, and social issues related to local genetic services.

[10] *Council of Regional . . .*

Reimbursement for Genetic Counseling, Testing and Newborn Screening

The issue of reimbursement for genetic counseling and genetic tests, including the genetic screening provided by the OSDH newborn screening program, was identified as a potential barrier to genetic services during the genetics needs assessment conducted by OSDH in June 2001. This issue has been recognized as a problem nationally. Genetic counseling is an essential service to ensure individuals and families understand the implications and limitations of genetic testing. In most cases, genetic testing does not predict absolutely whether an individual will manifest a condition later in life, with the rare exception of single gene disorders such as Huntington disease. Rather, a positive result indicates the increased likelihood of illness based on an individual's genetic makeup. Board-certified genetic counselors are individuals trained to effectively communicate information about the heritability of disorder/disease, available genetic testing, screening, prognosis, and recurrence within a framework of understanding and support. However, access to a board-certified counselor is limited to the physicians who are committed to providing genetic counseling despite the lack of reimbursement for such services. In the United States, there are no state or federal laws requiring insurers to offer or cover genetic counseling (Johnson 2).[27]

For genetic testing, local genetic laboratories reported insurance providers often do not acknowledge them as a reimbursable provider. Therefore, the geneticist is often forced to utilize an out-of-state lab, unless the patient is willing to pay for the genetic test. This can result in delayed receipt of test results, and can inhibit consultation between ordering geneticists and the laboratory clinical experts, which is necessary to facilitate the diagnostic process. For the public health newborn screening program, the fee has been inadequate to cover current testing and is the limiting step in the expansion of screening, such as Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD), that could reduce morbidity and mortality in children. The plan addresses issues of reimbursement through collaboration with OGAC and its committees, insurance providers, stakeholders, and the genetics program.

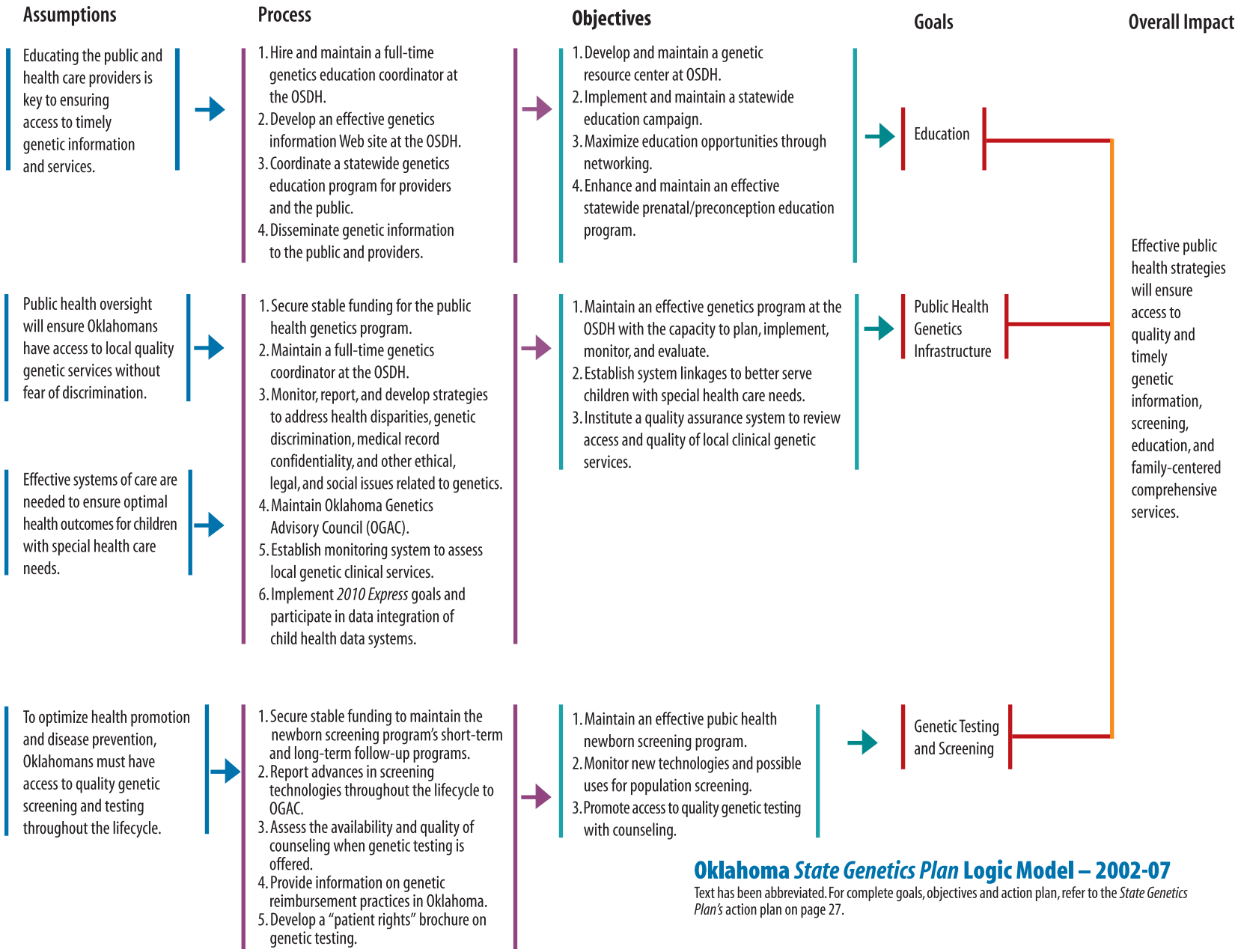
Evaluation

Evaluation will be an essential component of the public health genetics program. To determine if the OSDH genetics program is achieving the mission of the state plan, a comprehensive assessment and evaluation system will be established. The Institute of Medicine recommends every public health agency regularly and systematically collect, assemble, analyze and make available information on the health of the community, including statistics on health status, community health needs and epidemiologic and other studies of health programs (Khoury and Genetics 1719).[29] A logic model (page 42) of the state genetic plan has been developed. Utilizing the logic model and with the collaboration of the Evaluation Committee of OGAC, a comprehensive evaluation plan will be established to monitor goals and mission. Performance measures and outcomes will be clearly defined and monitored. Mechanisms will be established to track whether or not planned project activities (process) were actually carried out. Standardized data tools will be developed and incorporated into current data assessment programs such as Pregnancy Risk Assessment Monitoring System (PRAMS), and The Oklahoma Toddler Survey (TOTS). PRAMS and TOTS are OSDH population-based surveillance systems that provide health data on women and toddlers in Oklahoma. Data tools will be developed for the six core outcomes of 2010 Express and for related Title V performance measures. Effectiveness of the attainment of goals and mission will be monitored and assessed by the genetics program in collaboration with OGAC. The *State Genetics Plan's* action steps will be tailored as needed to efficiently and effectively achieve the mission.

[27] *Johnson, Alissa. . .*

[29] *From Genes to Public Health. . .*

Genetics Program of the Oklahoma State Department of Health



Oklahoma *State Genetics Plan* Logic Model – 2002-07

Text has been abbreviated. For complete goals, objectives and action plan, refer to the *State Genetics Plan's* action plan on page 27.

Summary

Goal 2

Through participation of the Oklahoma Genetics Advisory Council (OGAC) and its committees a comprehensive representative action plan for goal 2 was developed. The *State Genetics Plan's* three objectives for goal 2 include: (1) public health genetic infrastructure development, (2) system linkages to better serve children with special health care needs (CSHCN) population, and (3) clinical genetic services. The combined Public Health Policy and Evaluation Committee of OGAC identified five action steps as priority for implementation.

Goal 2 Action Plan

"Develop and maintain a responsive public health genetics program to plan, implement, monitor, and evaluate genetics education and services in Oklahoma."

Objective 1- The OSDH will develop and sustain a responsive, collaborative, culturally sensitive, and effective public health genetics program that links with community partners and addresses health disparities.

Action Steps (◇ symbol identifies prioritized action steps):

- ◇ Identify stable funding sources for the public health genetics program (priority number 2).
- ◇ In collaboration with Oklahoma Genetics Advisory Council (OGAC), establish a network to address and monitor local ethical, legal, and social issues related to genetics including confidentiality and discrimination, e.g., life insurance discrimination based on genetic testing, inflated insurance premiums due to a diagnosis of a genetic disorder, or Medicaid eligibility (priority number 3).
- ○ Establish a system for the public to report incidents of insurance and employee discrimination related to genetics.
- ○ Maintain a full-time State Genetics Coordinator at the OSDH to provide oversight of the implementation of the *State Genetics Plan* and to administer the public health genetics program.

- ○ Maintain the genetics advisory council (Oklahoma Genetics Advisory Council) as a diverse and active advisory group involved in strategic planning and evaluation of genetic programs.
- ○ Provide annual presentations on cultural diversity to OGAC.
- ○ Collaborate with the Family Advisory Committee of OGAC to ensure genetics services and information are culturally sensitive.
- ○ Establish a system to monitor and evaluate program performance.
- ○ Addition of members representing minority cultures to OGAC and its committees, i.e., Native American, African Americans, Hispanic, and Asian.
- ○ Through education and collaboration with community partners, provide public health leadership to promote quality genetics services in Oklahoma that are accessible and culturally sensitive.
- ○ In collaboration with the Public Health Policy Committee of OGAC, develop strategies to improve reimbursement for genetic services.
- ○ In collaboration with the Public Health Policy Committee of OGAC, determine an appropriate organizational structure for a public health genetics program within OSDH, including potential staffing and financial needs.
- ○ Network with national organizations to assist with program development for quality genetic services in Oklahoma, i.e., Genetic Alliance, Coalition of State Genetics Coordinators, International Society of Nurses in Genetics, National Newborn Screening and Genetics Resource Center, CDC Office of Genetics and Disease Prevention, American Society of Human Genetics, National Society of Genetic Counselors, and American College of Human Genetics.
- ○ Interface with public health programs (i.e., newborn screening and birth defects registry, Chronic Disease Service, Women's Health Service etc.), county health departments, and Children with Special Health Care Needs of Department of Human Services (DHS) to integrate genetics into these programs and provide education to program staff to improve surveillance and referral to clinical genetic services.
- ○ Establish relationships with programs that have not used genetic tools in the past, i.e., Chronic Disease, Acute Disease, and Environmental Health.
- ○ Collaborate with the State Epidemiologist on potential opportunities for local genetic epidemiological studies.

Objective 2 - The OSDH Genetics Program will establish system linkages to better serve children with special health care needs.

Action Steps (◇ symbol identifies prioritized action steps):

- ◇ Develop a system for integration of public health services through system linkages and adequate genetic program resources to ensure children with special health care needs are met (priority number 1).
- ◇ In collaboration with the Newborn Screening Programs and Pediatric Committee of OGAC, assure the medical home includes access to pediatric sub-specialty care (priority number 5).
- ○ Develop and promote the Family Health Services data integration plans to improve identification of children in need of genetic services by:
  - i. Continuing to implement the data integration of the Newborn Metabolic Disorder Screening Program and Newborn Hearing Screening Program, and Vital Records.
  - ii. Reviewing the feasibility of linking the newborn screening programs database with the Oklahoma State Immunization Information System (OSIIS).
  - iii. Reviewing the feasibility of linking the newborn screening programs database with the WIC and Lead Screening programs.
  - iv. Developing a data system for the long-term follow-up, to include links to the pediatric sub-specialist, of infants identified with a disorder through newborn screening.
  - v. Reviewing the feasibility of Web-enabled data system to improve follow-up of CSHCN to include linking and sharing information with the pediatric sub-specialist and medical home

- ○ Collaborate with the OSDH HIPPA representative to review access and release of information related to data integration efforts and program data, including newborn screening.
- ○ Promote a system of communication between genetic providers, pediatric sub-specialist, and the medical home to facilitate and ensure the needs of the child and family are met.
- ○ Collaborate and assure program linkages exist with Family Voices, Early Intervention, and CSHCN to ensure families of children with special health care needs are met.
- ○ Maintain CSHCN representatives (families and professionals) and other public health programs that serve CSHCN on the OGAC and Newborn Screening Programs and Pediatric Committee of OGAC.
- ○ Establish monthly or quarterly meetings between OSDH genetics program and the Department of Human Services CSHCN staff to identify opportunities to collaborate and share data on CSHCN population.
- ○ Collaborate with the Birth Defects Registry, Prenatal Screening and Diagnosis Committee of OGAC in assessing the feasibility of utilizing the birth defects registry to identify populations who would benefit from genetic services and establish a system to notify parents.

Objective 3 - The OSDH Genetics Program, in collaboration with OGAC and its committees, will assure that clinical genetic services are of the highest quality.

Action Steps (◇ symbol identifies prioritized action steps):

- ◇ In collaboration with the Evaluation Committee of OGAC, establish a system at the OSDH to monitor availability, quality, utilization, and accessibility of genetic clinical services, to include monitoring for health disparities (priority number 4).
- ○ In collaboration with the Public Health Policy Committee of OGAC, develop recommendations (standards) for:
  - i. quality genetic clinical services, including culturally sensitive issues, i.e., services are provided that are sensitive to the culture of the consumer.
  - ii. indications for referral to genetic clinical services