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...*