

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 20<sup>th</sup> 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, Turner's 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*).<sup>47</sup>

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.

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<sup>47</sup> *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.

