National Institutes of Health





Fact Sheet

Genes: What We Knew, Know, and Hope to Learn

Yesterday

- Scientists knew the structure of DNA and that genes code for proteins, but they did not recognize the complexity of how genes are regulated.
- The molecular biology revolution was in its infancy. The first few human genes were cloned, and DNA sequencing was just being developed.
- Efforts were just beginning to identify and map genes at the molecular level and to correlate genes with diseases. Only a few human diseases, such as sickle cell disease, were associated with abnormal genes.

Today

- The entire human genome and the genomes of hundreds of other species have been sequenced, providing a valuable resource for current and future biomedical advances.
- We know that species ranging from microbes to humans have similar genes and genetic pathways.
 By studying the genes of model organisms and computationally comparing the DNA sequences of different species, researchers discovered the functions of many human genes, including those associated with certain diseases.
- Computational tools allow scientists to analyze how patterns of gene activity are involved in health and disease. The activity patterns of hundreds of genes often reveal more about an organism than does the activity of individual genes.
- The information contained in gene sequences is far more complex than anyone imagined. For example, scientists learned that genes contain large regions of non-coding DNA that regulate gene activity. They also discovered that the body can read the same DNA sequence in different ways to produce different proteins.

- Scientists now know that gene regulation is central to an organism's development and to a cell's identity and function.
- Researchers are finding that the regulation of gene expression is highly complex and depends upon specific proteins binding to the DNA. These proteins, which include transcription factors and repressors, detect the cell's ever-changing needs and respond by regulating the activity of certain genes.
- Scientists continue to discover new roles for RNA, a
 class of molecule once thought to serve primarily as
 an intermediate between DNA and proteins during
 protein synthesis. They now know that RNA
 molecules can regulate gene expression and catalyze
 enzymatic reactions, including those that link amino
 acids together to form proteins.
- As scientists have known for more than 100 years, inherited traits are passed from parent to child through genes. But there is a growing appreciation for the many ways not directly tied to DNA sequence that traits can be inherited. These so-called epigenetic processes can profoundly impact the inheritance of traits, including those related to health and disease.
- Genetic tools—such as DNA fingerprinting—are
 used for a wide variety of purposes, including in
 criminal forensics, paternity testing, identifying
 human remains, matching organ donors, studying
 the inheritance of specific traits in human and animal
 populations, and understanding the migration
 patterns of ancient peoples.
- Genetics-based biotechnology has grown into a multibillion-dollar industry. It has created new drugs to treat conditions such as diabetes, anemia, growth hormone deficiency, and certain cancers and infectious diseases. It has also led to tests that diagnose rare diseases and predict a person's risk for more common diseases, such as breast cancer.

• Biotechnology is creating new research tools to fuel the next generation of discoveries.

Tomorrow

- Through improved methods to identify genetic risk factors for common, complex diseases like diabetes, heart disease, and cancer, doctors will be better able to prevent, predict, and treat these diseases.
- Doctors will use genetic information to tailor drug prescriptions, screening tests, and lifestyle recommendations for each patient.
- Through genetic studies of bacteria, viruses, and other organisms, researchers will better understand how microbes evolve. This will enable them to develop tools to predict and prevent the emergence and spread of infectious diseases.

- Using integrative approaches that simultaneously analyze many biological processes, scientists will learn how genes and the environment interact to contribute to disease.
- Advances in manipulating genes and producing proteins under controlled conditions will improve our ability to produce certain pharmaceuticals, foods, industrial chemicals, agricultural products, and biotechnology tools.

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