II. A Brief Overview of Genetics and Genetic Research

Human beings contain tens of thousands of genes, the basic material for cell function including the transmission of hereditary characteristics. Genes play a part in shaping many human characteristics, from the color of our hair and eyes to our level of risk for contracting a variety of diseases and conditions including depression, nearsightedness, allergies, and cancer.

Information about genes is stored in DNA (deoxyribonucleic acid). DNA is composed of four chemical bases: adenine, guanine, cytosine, and thymine, which appear as pairs (adenine with thymine, and guanine with cytosine) in the DNA. In normal conditions, each gene is carried in two copies, one inherited from each parent. Genes occur in particular locations along one of forty-six chromosomes, which, like genes, come half from the mother and half from the father. Each chromosome contains many thousands of genes and many millions of base pairs. The human genome, which encompasses all of the genetic information in the human body, consists of about three billion base pairs. Decoding a particular gene means learning the order of base pairs in that gene. Genes vary in size according to the number of base pairs of which they are comprised.

Genes control the functioning of the human body by producing proteins through processes called transcription and translation. Different proteins perform different functions. For example, some genes produce proteins like hemoglobin, which carries oxygen around the body. Other genes produce collagen, which provides the body with structural support. Still others produce enzymes that facilitate chemical reactions like digestion. Genes called tumor suppressors produce proteins that control or regulate cell
division. If cell division is not properly regulated, cells may divide too much or too rapidly and cancer may result.

Ordinarily, when new cells are produced through cell division, the DNA from the prototypical cell is replicated in the new cells. Sometimes, however, an error in the replication process may result in a change in genetic material, called a mutation. Mutations may also result from the exposure of a cell to radiation or chemical toxins. Mutations may be an alteration in the larger structure of the chromosomes or in specific base pairs, either through the deletion or reordering of one or more base pairs. Some mutations are advantageous to the cell or have little effect. Others, referred to as deleterious mutations, are harmful, causing genes to produce partially or completely non-functional proteins. While mutations in all cells can affect an individual’s health, only mutations in sex cells, called germline mutations, will be passed on to one’s offspring. Mutations in other cells, called somatic mutations, will not be inherited.

Mutations are actually quite common. It is believed that every individual is born with between five and thirty significant germline mutations in our DNA. Each may affect the risk for any of thousands of disorders, from allergies to depression to diabetes, that have been linked to heredity. Some hereditary disorders are caused by an inherited mutation in a single gene. These disorders, often referred to as Mendelian or single-gene disorders, are characterized as either dominant or recessive. A dominant Mendelian disorder requires a deleterious mutation in only one of the two copies of a gene that an individual carries. An example is Huntington’s disease. A recessive Mendelian disorder requires a deleterious mutation in both copies of a gene that an individual carries. Individuals carrying only a single mutation are known as trait carriers, and usually exhibit
no harmful effects. An example is Tay-Sachs disease. Over the past thirty years genetic research has yielded the gene responsible for many of these disorders, including sickle cell disease, Duchenne muscular dystrophy, and Huntington’s disease.

Many common diseases, such as cancers, diabetes, and hypertension, also have a genetic basis. However, these diseases are often not inherited in a direct Mendelian fashion. Instead, they develop as the result of complex interactions of genes and environmental stimuli, such as chemical toxins, diet, and lifestyle. In conducting genetic research on such multifactoral diseases, scientists aim to identify and understand the roles of genes in the onset and course of the disease so that diagnostic tests and eventually improved clinical treatments may be developed.

Initial genetic research on a disease often looks to identify genes that increase an individual’s risk for the disease. To look for such genes geneticists commonly use association studies. Association studies examine whether variations in certain genetic base pairs correspond to disease symptoms. If this correspondence occurs at a rate above that predicted by chance alone, it is likely that the base pairs are linked to a gene that affects risk for the disease. However, for this to be confirmed, geneticists must conduct additional studies to code the gene and demonstrate that it affects disease risk in large samples of people.

Once a gene is identified additional genetic research is needed to understand its role in the onset and course of the disease. This research includes studies to identify specific mutations in the gene and estimate their distribution and the levels of risk they confer. It also includes studies to understand the function of the gene and how this function affects other genes or environmental factors that may contribute to the onset of
the disease or the types of symptoms it presents. Studies may also be conducted to
determine whether the gene has an affect on other diseases. Ultimately, if gene mutations
are discovered that confer a significant level of risk for the disease, it may be possible to
develop diagnostic tests to identify individuals at elevated risk. Such tests can allow high
risk individuals to make dietary or lifestyle changes to decrease their risk or in some
cases undergo preventive procedures or drug regiments. Similarly, genetic research that
identifies genes that affect the body’s response to different drugs can lead to the more
efficient prescription of medications. As of yet, genetic therapies that can correct
deleterious mutations carried by an individual have not been developed, although some
researchers hope that such therapies will become available in the future.

While genes explain much about the way individuals live and function, all too
often both physicians and lay people may be guilty of “genetic essentialism,” a tendency
to view people as solely a creation of their genes. Human beings are more than genetics.
To reduce everything to genes and mutations is to minimize other traits of humanity as
well as the possibilities for improving the quality of our lives through manipulations of
diet, workplace, and environment. We are more than the sum total of our genes.

Readings

Adelman (2006) contains a discussion of “hows” and “whys” of the decoding of the
genome.
Begley, Hayden, Underhill, and Beals (2000) provide a brief introduction to the Human
Genome Project.
Cowley, Underwood, and Check (2000) discuss how genetic science is transforming both
medical research and medical practice.
NIH, National Human Genome Research Institute, Educational Resources
[http://www.genome.gov/Education/] presents a user friendly guide to human
genetics and the Human Genome Project.
U.S. Dept. of Energy Office of Science, Human Genome Program, Human Genome
Project Information
[http://www.ornl.gov/sci/techresources/Human_Genome/home.shtml] provides a
concise overview of human genetics and an introduction to the Human Genome Project. It is also linked to “Gene Gateway,” which is a non-technical guide to other genome resources on the web.


World Health Organization, The Genomic Resource Centre [http://www.who.int/genomics/en/] contains extensive information on genetics geared toward policymakers and the public, including an international database of documents on the ethical, legal, and social issues raised by genetics.