

Fact Sheet on Medical Genetics

Genetics is the study of individual genes and their effects.

Medical Genetics is any application of genetic principles to medical practice. This includes studies of inheritance, mapping disease genes, diagnosis and treatment, and genetic counseling.

Genomic medicine is the study of conditions that are partly caused or prevented by mutation(s) in gene(s). Genomics is the study not just of single genes, but also of the functions and interactions of all the genes in the genome. These complex processes are the genetic causes of many common diseases, such as asthma, hypertension, diabetes and psychiatric disorders.

Genetic Counseling is a communication process that deals with the human problems associated with the occurrence, or the risk of occurrence, of a genetic disorder in a family. The goals are to assist the family in understanding:

- The diagnosis and implications of a condition
- The role of heredity
- Recurrence risks and options
- Possible courses of action
- Methods of on-going adjustment

DNA (deoxyribonucleic acid) - The molecule inside each cell that carries the genetic instructions for making living organisms. DNA is double-stranded and made up of four building blocks called nucleotide bases (different chemicals that are abbreviated A, T, C, and G) that are repeated over and over throughout a genome. The human genome has 3 billion pairs of bases. The order of these nucleotides contains the instructions in the gene to tell the cell how to make a particular protein.

Gene - The functional and physical unit of heredity passed from parent to offspring. Genes are pieces of DNA and carry information for making all the proteins required by all organisms. These proteins determine, among other things, how the organism looks, how well its body metabolizes food or fights infection, and sometimes even how it behaves.

The number of human genes is about 30,000. Each gene can vary in length and cover thousands of bases.

Genome - All the DNA contained in an organism or a cell. The genome consists of about 3-4 billion base pairs. The action of much of the genome is unknown.

Mutation - A permanent structural alteration or change in DNA. While some mutations are harmful, in most cases DNA changes either have no effect or cause harm, but occasionally a mutation can improve an organism's chance of surviving and the beneficial change can be passed on to its descendants.

What is the impact of genetics on health and disease?

- It is estimated that 3-7% of the general population will be diagnosed with a recognized genetic disorder, NOT INCLUDING common disorders, such as cancer, diabetes, heart disease and psychiatric disorders.
- All disease (with the possible exception of trauma) is genetic.
- Understanding how variations in an individual's DNA may affect disease and health is the focus of genomic medicine. This knowledge can lead to
 - innovative ways to diagnose disease
 - earlier detection by being able to identify genetic predisposition to a particular disease
 - new approaches to treatment
 - development of specific designer drugs that target a mutation [pharmacogenomics]

Genetic Testing includes methods to look for changes or abnormalities in an individual's genes or in the gene products (e.g., proteins or the chemicals the genes make or regulate). The purpose is to determine if someone has a genetic condition or is likely to get a specific disorder. Typically a person may be offered testing if they have a family history of a specific disease; they have symptoms of a genetic disorder, or are concerned about passing a genetic disorder to their offspring. Genetic testing is voluntary, as the testing has both benefits and limitations.

Ethical, Legal and Social Issues (ELSI) of the Human Genome Project (HGP) and Genetic Testing: Information that is available from genetic testing and from the knowledge gained by the HGP has great potential to improve health but also may raise concerns for individuals, family members and society as a whole. Some of these include: How should the information be used? Who should have access to the information (privacy; informed consent)? How can we be protected from harm from improper use or disclosure (confidentiality)? A certain proportion of research dollars that the government earmarks for the Human Genome Project must be spent on addressing these issues, amounting to more than \$14 million annually.