

# THE HUMAN GENOME - THE BLUEPRINT OF LIFE

The Human Genome project sequenced DNA, the molecules that make up chromosomes in cells. The information derived from this project presented scientists with a valuable opportunity to not only uncover the secrets of DNA but also the manner in which genes are associated with disease. Scientists now are able to compare the genomes of people who have a certain condition with those who do not, in order to determine whether genetic variation plays a role in that condition. This information will help them to predict and possibly prevent disease in the future.

## 1. Cell

Each of the billions of cells in the human body contains all chromosomes packed tightly into the region called the nucleus.

## 2. Chromosomes

Half of the chromosomes in the nucleus come from your mother, and half from your father. Each chromosome is a long, tightly coiled molecule called DNA, or deoxyribonucleic acid.

## 3. DNA

If uncoiled, the DNA from all the chromosomes in a single cell placed end to end would stretch more than six feet.

## 4. Genome

DNA is made up of chemical building blocks abbreviated A, C, T, and G. The entire length of a DNA strand consists of these four blocks in different combinations. Together, all the DNA in all the chromosomes - more than 3 billion letters - makes up the human genome. When scientists say they have "sequenced" the human genome, they mean that they have figured out the order of all those A's, C's, T's, and G's in sequence.

## 5. Genes: 30,000 DNA Segments

Much of the DNA in the genome is organized into units called genes. There may be as many as 30,000 genes in the genome; they are the instruction manual for making all the proteins in the body. These proteins are the physical "stuff" that makes up our hair, skin, teeth, and bones, among other things. They also control chemical reactions, regulate blood sugar and heart rate, and control how food in the digestive tract is metabolized in the body.

## 6. Misspellings in the Sequence

The way the genes are "read" makes all the difference - one letter out of place in a gene can cause disease. Now that we have the normal sequence of the human genome, researchers can compare the DNA sequences from people who have a disease or condition to those who don't. If there are differences in the spelling of certain genes between the two groups, it's possible that the condition may be caused by or related to that misspelling in that gene.

## 7. Genes and Disease

Scientists have identified about 6,000 diseases, such as Huntington disease and cystic fibrosis, that are directly caused by misspellings or unusual problems in single genes. But the genetic contribution to many common conditions - such as diabetes and heart disease - is part of a larger puzzle that could include diet, lifestyle, environment, and even other genes. For many of these common conditions, genetic misspellings probably make only a small contribution to disease relative to other factors, or work in concert with them to cause illness.

# Elements Of Human Genetics

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