

# Genetic screening

Many people have heard, whether on television, from a newspaper, a magazine, or on the radio about the discovery of a gene for a certain disease. What does this really mean? For several years scientists have been trying to understand how some diseases are inherited and what the underlying cause of certain fatal diseases are. The term "genetic" refers to traits which are inherited from our parents.

## Chromosomes

Inside our body's cells, in the nucleus, are our chromosomes (they look like little sticks). Each of us has 46 chromosomes, which come in pairs of twenty-three. We can organize each pair by size and appearance because each chromosome pair looks different. For instance, some chromosomes are very large and others are very small. The first twenty-two pairs are the same in men and women, but the 23rd pair is different between the sexes. We call this last pair our sex chromosomes and we identify them with letters instead of numbers (i.e., women have two X chromosomes and men have one X and one Y chromosome). Therefore, if we were to look at the chromosomes in someone's cells, we could determine if that person was a male or female by just looking at their sex chromosomes.

## Genes

What makes the chromosomes so important is that they contain our genes. We receive all the instructions as to how we will develop from our genes. On each chromosome there are hundreds to thousands of genes (our larger chromosomes have more genes than our smaller ones). Each gene has a specific function for our bodies, such as determining our eye color or making a particular protein. Collectively, all of our genes will determine how we will look, how our organs will develop, and how the chemicals in our bodies will form. DNA

Genes are made of DNA and DNA is made up of four biochemical substances: adenine, guanine, cytosine, and thymine (A, G, C, and T). The combination of these substances determine the DNA sequence, which is specific for each gene. We have so many different genes because there are practically an infinite number of combinations of these four substances. If there is a change in one of these chemicals, such as an A replacing a C, this could prevent the gene from functioning. This change is called a mutation. If a gene cannot function, this can cause a genetic disease for an individual.

## How are we affected?

For many diseases you must have two non-working genes to have a problem. This means that on one chromosome there is a specific gene that is not working because of a mutation, and on the other matching chromosome, there is another gene with a mutation. Therefore, since both copies of the gene are not working, this individual will have a certain disease because the body is missing an important chemical. On the other hand, if a person has only one chromosome with a non-working gene, and the other matching chromosome has a working gene, this person will not have the disease because the working gene can compensate for the gene with the mutation. People who contain one non-working gene are known as **carriers** and these people are healthy.

## What does it mean to be a carrier?

All humans have 5-8 genes that are not working. This means that we are all carriers for at least 5 genetic diseases. **Genetic screening refers to the ability to detect mutations in a person's DNA to determine if they are a carrier for a particular disease.** If we know the gene for a specific disease, and if we can identify the mutations that cause the disease, then we can examine a person's DNA and determine if they have a mutation on one of their genes. If a person learns that he or she is a carrier, they just happen to know one of the 5-8 genetic diseases they carry a gene for.

## Genetic screening based on ethnicity

There are **thousands** of diseases which result from genes that are not working properly. Some diseases are more common in certain ethnic groups. For instance, in the Caucasian population, cystic fibrosis (CF) is the most common genetic disease. In the African-American population, sickle cell is seen most frequently, and in the Jewish population, Tay-Sachs has a higher incidence rate. However, it's important to understand that even though some diseases are common to certain ethnic groups, no group is immune. In other words, even though CF is most commonly seen in the Caucasian population, it **does** appear in

individuals from other ethnic backgrounds, just not as frequently.

For many of these conditions, testing is available to determine one's carrier status. For those couples who discover that **both** members are carriers, prenatal diagnosis may be an option. This issue can be addressed with your physician and/or genetic counselor.