What is Genetic Testing?

During the last 10 years, scientists have learned many new facts about the role of genes in common diseases. For example, changes, or mutations, in certain genes can increase a person's risk for getting a disease such as cancer. Tests are now available to identify a few disease-related genes. Genetic testing involves looking for these changed genes in a sample of blood or tissue.

What are genes?

Each cell in the human body has 23 pairs of chromosomes. One set of chromosomes comes from a person's mother and the other set from the father. Each chromosome is made up of smaller units called genes (See Figure 1). As beads are linked to make a necklace, genes are linked to make a chromosome. Genes carry the information or blueprint for who and what a person is. Different genes are turned on in different cells to give each type of cell (for example, a brain cell or a muscle cell) its own makeup. When genes change, or mutate, this may cause disease.

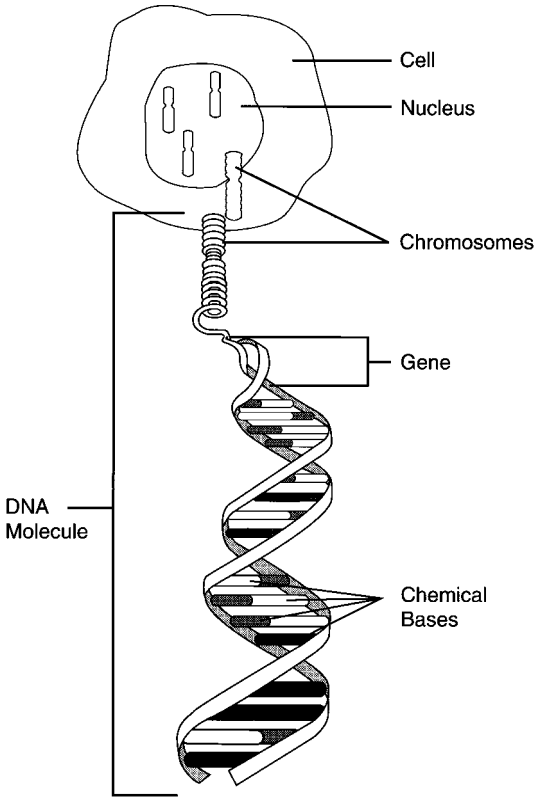

Figure 1

DNA carries the instructions that allow cells to make proteins. DNA is packaged in units called chromosomes, housed in the cell's nucleus. Genes are working sub-units of DNA.

What is the relationship between genes and cancer?

About 5 to 10 percent of all cancers are hereditary. This means that a gene change or mutation that increases the risk for cancer is inherited from a parent. Although people who inherit a gene change face a greater chance of having cancer, not all will develop the disease. Scientists have identified certain cancer-related genes from studying families who report a higher than average number of cancers among their relatives. For example, changes in the BRCA1 (Breast Cancer 1) and BRCA2 (Breast Cancer 2) genes are known to increase a person’s risk of developing cancers of the breast and ovaries. Another gene, the APC gene, is linked to a type of colon cancer known as familial adenomatous polyposis.

What is the role of genetic counseling?

Genetic testing is available for several hereditary cancer genes. In many cases, genetic testing is recommended only for individuals who have a strong family history of cancer. Persons who undergo genetic testing are often asked to provide a complete history of all cancers in their

family. A pedigree or family tree is often used to show the family history. The risk of having a hereditary form of cancer is evaluated from this information.

Before testing, it is important that people fully understand the potential benefits and limitations of testing, as well as the possible implications for themselves and their families. People who consider genetic testing will usually first have genetic counseling. The process of genetic testing and its risks and benefits are explained during one or more visits.

If you choose to have genetic testing, you will likely have genetic counseling when you receive your test results. During this session, the test results are explained and follow-up recommendations are made for you and your family.

What are the benefits of testing?

Genetic testing may be used to help identify people who are at high risk for developing certain types of cancer before any symptoms appear.

Because these forms of cancer often occur earlier in life than expected, doctors may recommend that people with a high cancer risk begin routine screening at a younger age than what is usually advised for the general population. High-risk screening enables health professionals to detect cancers as early as possible, when there is the best chance of successful treatment and cure.

Genetic testing also may be used to identify members in a high-risk family who do not carry a cancer-related gene change and thus do not need to undergo early cancer screening. However, like the general population, these individuals are still at risk for developing cancer and still need to follow general cancer screening guidelines.

There may also be emotional benefits of having genetic testing. Some individuals report a sense of relief and a feeling of control from learning whether or not they are at high-risk of developing cancer. Genetic testing may also resolve feelings of uncertainty about one's own future or the future of one's children. Results of genetic testing can enable one to make informed decisions about future health care needs. On the other hand, some people report that they would feel anxious if they knew they carried a cancer-related gene change.

What are the limitations?

Although a positive test result can tell you if you carry a mutation, it cannot tell you when or if cancer will develop. Another unknown factor is the effect of the environment (for example, air pollution or chemical exposure) on these genes.

Remember, some people with a gene change may **never** develop cancer. At present, there is no way to completely prevent cancer from developing.

Because genetic testing is so new, when a test result is negative, it is possible that a gene change was missed or that other genes that have not yet been discovered may be involved. Also, a negative test result **does not** mean that you won't get cancer. A person who does not inherit a gene change has a risk of developing cancer equal to the risk found in the general population. Thus, it is still important to follow routine screening guidelines for the detection of cancer.

In general, genetic testing involves many ethical, legal, and social issues. Genetic discrimination may be a problem despite the lobbying of the medical community and the passage of several laws to protect families. There is currently legislation on the state and national level to protect individuals in group health plans from discrimination based on genetic testing results. Your genetic counselor will give you information regarding this issue before you undergo testing.

The expense of testing is another issue you must consider. Some insurance companies will not pay for genetic testing. If you choose to pay for it yourself, the cost can be high.

You must also consider how genetic testing may cause changes in family relationships. Some family members may wish to be tested and know the results; others may prefer not to participate at all.

Think about all the issues described above and discuss them with your health care team **before** you decide to have genetic testing.

Where can I find more information?

If you have questions about testing or specific types of cancer, ask your doctor. If you wish to have your risk evaluated because several members of your family have cancer, ask for an appointment in one of the risk assessment clinics at M. D. Anderson Cancer Center.

For Information on Genetic Testing

National Cancer Institute (NCI)
1-800-4-CANCER (1-800-422-6237)
http://www.nci.nih.gov.