

## What are genes?

Every cell in your body contains genetic material, or *genes*. Genes are the blueprints for your body. They determine what color eyes you have and how tall you are. They also affect other functions of your body. For example, they tell your body to repair tissue that has been injured and help a woman's body prepare for a growing baby during pregnancy. Sometimes though, your genes do not work like they should. This is due to an error in one or more of your genes, called a *mutation*.

Mutations may be inherited or spontaneous. Inherited mutations are those you were born with — a defective gene that one of your parents passed on to you at birth. Spontaneous mutations are those that may occur in a single cell during the course of your life. There are many ways a spontaneous mutation can happen. However, scientists do not yet know exactly how, or if, these mutations are related to a woman's lifestyle (diet, exercise and reproductive patterns), chemical changes inside the body or exposure to environmental toxins such as radiation or chemicals — or if these mutations can even be prevented.

## Genes and breast cancer

Scientists have found two specific genes that when mutated are important in the development of breast cancer. They are called BRCA1 and BRCA2. Every woman has these genes, but some women have inherited a mutated form of one or both genes. Inheriting a mutated form of BRCA1 or BRCA2 increases a woman's risk of breast and ovarian cancer. However, not all breast cancers are due to inherited mutations.

Inherited gene mutations, including mutations in BRCA1 and BRCA2, account for only about 5 to 10 percent of all cases of breast cancer,<sup>1</sup> while most breast cancers are due to spontaneous gene mutations.

## Who has mutations in BRCA1 and BRCA2?

The likelihood that you have mutations in the BRCA1 or BRCA2 genes is greater if one or more of the following statements is true for you:

- you are younger and have been diagnosed with breast cancer (below age 50)
- your mother, sister or daughter has had breast cancer before age 50 or ovarian cancer at any age
- a woman in your family has had both breast cancer and ovarian cancer
- a woman in your family has had breast cancer in both breasts
- your family is of Ashkenazi Jewish descent
- a male in your family has had breast cancer

Remember, most women who get breast cancer do not have an inherited gene mutation in BRCA1 or BRCA2. All women should be screened with routine mammograms and clinical breast exams.

Mutations in the BRCA genes are not only found in women. Men can also carry the abnormal genes, which may increase their risk of prostate cancer. Men with a BRCA2 mutation also have an increased risk of breast cancer.

<sup>1</sup> American Cancer Society, Breast Cancer Facts & Figures 2007-2008.

## Can I find out if I have an inherited gene mutation?

Yes, you can. Women who have a family history of breast cancer and are interested in being tested for an inherited gene mutation should seek a referral to a genetic counselor. Genetic counselors are trained health professionals who can interpret a woman's family health history as well as the results of genetic testing. The steps a woman can expect to go through with the genetic counselor are listed below:

**STEP 1:** You will provide a thorough family health history and the counselor will explain your individual risk.

**STEP 2:** Pre-test counseling will be held to help you decide whether or not to proceed with genetic testing. This counseling includes:

- an overview of the genetic test procedure
- a review of the risks and benefits of genetic testing, such as cost, privacy and the potential knowledge that you carry the gene mutation
- a discussion of what you will do with the information once you know the test result

- a discussion of the emotional impact of this information, as well as implications for your family

**STEP 3:** A sample of your blood will be drawn for the test if you decide to proceed with testing.

**STEP 4:** The sample will be sent for testing. It usually takes 3 weeks to obtain results.

**STEP 5:** Interpretation of the results will be explained to you by the genetic counselor.

Some people are concerned about being treated unfairly based on the result of a genetic test. State and federal laws protect you, including the Genetic Information Nondiscrimination Act (GINA) which will go into effect May 2009. GINA prevents health insurers from denying coverage or charging higher premiums for a person with an increased genetic risk of breast cancer. GINA also protects employees from unfair treatment in the workplace as a result of genetic testing.

## Where can I get genetic testing?

If you are interested in genetic testing, you should talk to your doctor. Your doctor can refer you to a genetic counselor if one is available in your area. If your doctor is not aware of a genetic counselor close to you, contact the National Cancer Institute or the National Society of Genetic Counselors. They can give you a referral to the health centers nearest you with genetic counselors on staff. They can also provide additional information about BRCA1, BRCA2 and genetic testing. These organizations may be able to provide additional information:

Susan G. Komen for the Cure®  
1-877 GO KOMEN (1-877-465-6636),  
[www.komen.org](http://www.komen.org)

American Cancer Society  
1-800-ACS-2345, [www.cancer.org](http://www.cancer.org)

Facing Our Risk of Cancer Empowered, Inc. (FORCE)  
1-866-824-7475, [www.facingourrisk.org](http://www.facingourrisk.org)

National Cancer Institute  
1-800-4-CANCER, [www.cancer.gov](http://www.cancer.gov)

National Society of Genetic Counselors, Inc  
1-312-321-5163, [www.nsgc.org](http://www.nsgc.org)

### Related fact sheets in this series:

- Breast Cancer Risk Factors
- Types of Breast Cancer
- What is Breast Cancer?