

What are genes?

Every cell in your body contains *genes*. Genes are the blueprints for your body. For example, they decide the color of your eyes. They also affect other functions of your body, such as when cells grow, divide and die. Sometimes, genes do not work like they should. This is due to an error in one or more genes, called a *mutation*.

Mutations may be inherited or spontaneous. Inherited are those you were born with. They are passed on to you by one of your parents. Spontaneous are those that may occur during your lifetime. There are many ways this can happen. We do not yet know how, or if, these are related to a woman's lifestyle (such as diet and exercise). Also, chemical changes inside the body and contact with environmental toxins such as radiation or chemicals are under study. We don't know if we can prevent these mutations.

Genes and breast cancer

The two most well-known genes linked to breast cancer are *BRCA1* and *BRCA2* (BReast CAncer genes 1 and 2). Everyone has these genes, but some have inherited a mutated form of one or both. Inheriting a mutated form of *BRCA1* or *BRCA2* increases a woman's risk of breast and ovarian cancer.

Most breast cancers are not linked to inherited mutations. Only about 5 to 10 percent of all cases of breast cancer in the United States are due to inherited gene mutations.

Who has mutations in *BRCA1* and *BRCA2*?

Most women who get breast cancer do not have an inherited *BRCA1* or *BRCA2* gene mutation. The chance that you have a *BRCA1* or *BRCA2* gene mutation is greater if one or more of these statements is true:

- you were diagnosed with breast cancer at an early age
- your mother, sister or daughter was diagnosed with breast cancer at an early age or ovarian cancer at any age
- a woman in your family has had breast 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

All women should be screened for breast cancer with routine mammograms and clinical breast exams.

What about Men?

BRCA gene mutations are not only found in women. Men can also carry these mutations and pass them on to their children. Men with a *BRCA2* mutation have an increased risk of breast and prostate cancer.

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

Yes, you can. Women who think they need to have genetic testing or have a strong family history of breast cancer should see a genetic counselor. This is a trained expert who can gather a woman's family health history. He or she can also explain the results of genetic tests. In most cases, the test is done first on the survivor. If no error is found, the cancer was not likely due to a mutation. So, no further testing of other family members is needed. The process:

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

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

- an overview of the 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. The Genetic Information Nondiscrimination Act (GINA) prevents health insurers from denying coverage or charging higher premiums for a person with an increased genetic risk of breast cancer. It also protects employees from unfair treatment at work.

Genetic testing costs

Check with your health insurance provider to find out whether the costs of genetic counseling and testing are covered in your plan. If you have a new insurance plan that began on or after August 1, 2012, the Affordable Care Act (health care reform) requires coverage of these costs (when recommended by a provider). If you have a *BRCA1* or *BRCA2* gene mutation, the Affordable Care Act also requires coverage of counseling to help you decide if taking medications to lower the risk of breast cancer is right for you.

Where can I get genetic testing?

If you would like to learn more about genetic testing, talk to your doctor. Your doctor can refer you to a genetic counselor in your area. If your doctor is not aware of one close to you, contact the National Cancer Institute or the National Society of Genetic Counselors. They can refer you to a center near you with counselors on staff. They can also provide more detail about *BRCA1*, *BRCA2* and genetic testing. These organizations may be able to provide additional information:

Susan G. Komen®
1-877 GO KOMEN (1-877-465-6636),
www.komen.org

American Cancer Society
1-800-ACS-2345, www.cancer.org

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

National Cancer Institute
1-800-4-CANCER, www.cancer.gov

National Society of Genetic Counselors, Inc
1-312-321-6834, www.nsgc.org

Related fact sheets in this series:

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

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