Genetics and Breast Cancer

Breast cancer is the most common cancer diagnosed in women after skin cancer and is the second leading cause of cancer deaths in women after lung cancer. Many women with breast cancer have family members with a history of breast cancer, and scientists believe that vulnerability to breast cancer sometimes has a **genetic** (inherited) component. **Mutations** (inherited genetic variations) of 2 **genes** (segments of DNA that are biological units of heredity) appear to account for about 5% of breast cancers diagnosed annually in the United States. The July 28, 2004, issue of *JAMA* includes an article about using a computer program to help patients make decisions about genetic testing for breast cancer.

BRCA1 and *BRCA2*

- ***BRCA1*** and ***BRCA2*** (**BR**east **CA**ncer **1** and **2**) are genes that have been discovered to play a role in some breast cancers.
- Most women have 2 normal copies of the *BRCA1* and *BRCA2* genes.
- An estimated 250,000 women in the United States have a mutation in one of these genes.
- Women in general have about a 12% chance (1 in 8 women) of developing breast cancer. Women with a *BRCA1* or a *BRCA2* mutation have up to an 87% lifetime risk of breast cancer. However, this means that at least 13% of women with these mutations will not develop breast cancer.
- Women with *BRCA1* or *BRCA2* mutations also have an increased lifetime risk of cancer of the ovaries—up to 54% for *BRCA1* and up to 27% for *BRCA2*.
- Women who test positive for *BRCA1* or *BRCA2* mutations should undergo further screening and take additional precautions, including frequent and early breast self-examinations, clinical breast examinations (performed by a doctor), and regular mammograms. They may consider other risk reduction options including the use of chemopreventive agents, such as tamoxifen, surgical removal of the ovaries, or surgical removal of the breasts.
- Men with a *BRCA2* mutation also have an increased risk of breast cancer—a 6% lifetime risk compared with the average lifetime risk of 0.1% in US men.

GENETIC TESTING

Genetic testing can determine whether a person has a specific genetic mutation that can increase the risk of certain diseases or disorders. Genetic tests can detect mutations in the *BRCA1* and *BRCA2* genes. Because most breast cancers are not caused by genetic mutations, genetic testing may only be of value if you believe you are at a high risk of having a *BRCA1* or *BRCA2* mutation. You are more likely to have these mutations if

- there are 3 or more women with breast cancer in a single generation of your family
- women develop breast cancer at a young age in your family (younger than 50 years)
- breast cancers in the family are often found in both breasts
- there are cases of breast and ovarian cancer in the same family

GENETIC COUNSELING

Genetic counseling provides individuals and families with information about the risks, benefits, and limitations of genetic testing, an assessment of the probability of carrying a genetic mutation, and the options to consider if a mutation is found.

FOR MORE INFORMATION

- American Cancer Society
  800/227-2345
  www.cancer.org
- National Cancer Institute
  800/4-CANCER (800/422-6237)
  www.cancer.gov
- Gilda's Club
  888/GILDA-4-U (888/445-3248)
  www.gildasclub.com

INFORM YOURSELF

To find this and other JAMA Patient Pages, go to the Patient Page link on *JAMA*'s Web site at www.jama.com. A Patient Page on cancer clinical trials was published in the June 9, 2004, issue; and one on preventing cancer was published in the May 26, 2004, issue.

Sources: National Cancer Institute, American Cancer Society, Gilda's Club, Susan G. Komen Breast Cancer Foundation

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