

Hereditary Breast and Ovarian Cancer Syndrome

Fact Sheet for Connecticut Healthcare Professionals

What is Hereditary Breast and Ovarian Cancer (HBOC) syndrome?

HBOC syndrome is a hereditary cancer predisposition condition usually caused by mutations in the *BRCA1* and *BRCA2* (breast cancer susceptibility genes 1 and 2). *BRCA1* and *BRCA2* belong to a class of genes known as tumor suppressors. In normal cells, *BRCA1* and *BRCA2* help ensure the stability of the cell's genetic material (DNA) and help prevent uncontrolled cell growth.

Facts about HBOC syndrome

- Approximately 5%-10% of breast cancer patients have HBOC syndrome.
- Approximately 10%-15% of ovarian cancer patients have HBOC syndrome.
- Women who inherit abnormal *BRCA1* and/or *BRCA2* genes have a 35%-80% risk of being diagnosed with breast cancer during their lifetimes, compared to a 12%-13% chance for women in the general population.
- Women who inherit abnormal *BRCA1* and/or *BRCA2* genes have a 40%-60% risk of being diagnosed with ovarian cancer during their lifetimes, compared to a 1%-2% chance for women in the general population. Although ovarian cancer is less common than breast cancer, it is more often fatal.
- Women with a harmful *BRCA* mutation are more likely than non-carriers to be diagnosed with cancer before age 50.
- Men with harmful *BRCA1* and/or *BRCA2* mutations have a 5%-10% risk of being diagnosed with breast cancer, compared to a 0.1% chance for men in the general population.

How common are *BRCA1* and *BRCA2* mutations?

- In the general population, between 1 in 300 and 1 in 800 individuals carries a *BRCA1* or *BRCA2* mutation.
- For individuals of Ashkenazi (Eastern European) Jewish ancestry, 1 in 40 individuals carries a *BRCA1* or *BRCA2* mutation.

Who is most likely to have a *BRCA1* or *BRCA2* mutation?

The likelihood is highest in families with a history of multiple cases of breast cancer, cases of both breast and ovarian cancer, one or more family members with two primary cancers (original tumors that develop at different sites in the body), or Ashkenazi Jewish ancestry. However, not every woman in such families carries a harmful *BRCA1* or *BRCA2* mutation, and not every cancer in such families is linked to a harmful mutation in one of these genes but instead may be the result of a sporadic mutation. Furthermore, not every woman who has a harmful *BRCA1* or *BRCA2* mutation will develop breast and/or ovarian cancer.

Specific indications for genetic counseling and testing vary among professional organizations. Guidelines are not a substitute for clinical judgment. Not all clinical scenarios can be anticipated, such as when there is a limited family structure or family medical history.

Why is genetic counseling important?

Genetic counseling helps people better understand their risk for hereditary cancer in order to make informed decisions about genetic testing and follow-up care.

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Connecticut Department of Public Health
410 Capitol Avenue, Hartford, CT 06106
(860) 509-8070 www.ct.gov/dph



What does genetic counseling entail?

Genetic counseling is a process that encompasses the following services:

- Reviewing an individual's personal and detailed family medical history
- Assessing and explaining risk for hereditary cancers and the chance of finding a mutation through genetic testing
- Discussing the benefits, limitations, and other possible consequences of genetic testing
- Outlining medical implications of a positive or a negative test result
- Determining which family member is most appropriate to begin the genetic testing process in a family
- Interpreting genetic test results and explaining what they mean for individuals and their relatives
- Providing referrals to experts for follow-up screening and risk management
- Providing referrals to support resources and research opportunities (including research on genetic testing, screening, treatment, etc.)
- Discussing risks and medical management options with a patient's other health care provider(s)
- Addressing common concerns about the privacy and confidentiality of personal genetic information

What are some of the benefits of genetic testing for breast and ovarian cancer risk?

- A positive test result can bring relief from uncertainty and allow people to make informed decisions about their futures. They can take steps to reduce their cancer risk through increased surveillance or other medical and lifestyle choices.
- A positive test result may help to explain why individuals or family members had cancer in the past, and, should they choose to share test results, may provide their family members with useful information.
- Those who have a positive test result may be able to participate in medical research that could, in the long run, help reduce deaths from breast cancer.
- A negative test result may provide a sense of relief and preclude the need for special preventive checkups, tests, or surgeries.

What are the disadvantages of genetic testing?

Test results may affect a person's emotions, family relationships, finances, privacy, and medical choices.

- A positive result may make a person feel anxious, angry, or depressed. Medical treatments, such as surgery to try to prevent the cancer, could have serious, long-term implications and uncertain effectiveness.
- A negative result may make a person feel guilty because they escaped a disease that affected a loved one. They may also get a false sense of security that they have no chance of getting cancer, when, in fact, their cancer risk is the same as that of the general population.
- Because genetic testing can reveal information about more than one family member, the emotions caused by the results can create tension within families. The results also can affect personal choices, such as marriage and childbearing.
- Privacy and confidentiality of genetic test results are additional potential concerns. There is no guarantee that a person's test results will remain private.
- Genetic testing can be expensive, costing about \$300 to \$4,000, depending on the extent of testing. Although many insurance plans cover the cost of the testing for women at high risk, coverage is unlikely for women not considered to be at high risk.