

Risk Assessment, Genetic Counseling, and Genetic Testing for BRCA-related Cancer in Women

The U.S. Preventive Services Task Force (Task Force) has issued a **final** recommendation statement on *Risk Assessment, Genetic Counseling, and Genetic Testing for BRCA-related Cancer in Women*.

This final recommendation statement applies to women who have no signs or symptoms of a BRCA-related cancer (cancer of the breast, ovary, fallopian tube, or peritoneum).

The Task Force reviewed recent research studies on risk assessment, genetic counseling, genetic testing, and treatments aimed at reducing the risk for BRCA-related cancers in women. The final recommendation statement summarizes what the Task Force learned about the potential benefits and harms of this multi-

step process: (1) Women who have family members with breast, ovarian, fallopian tube, or peritoneal cancer should be screened to see if their history is associated with an increased risk of carrying a BRCA mutation. If it is, women should get in-depth genetic counseling to review their family history and determine whether BRCA testing would be useful. If testing would be useful, women should receive genetic testing for BRCA mutations. (2) Women whose family history is not associated with increased risk for BRCA mutations should not have routine genetic counseling or BRCA testing.

This fact sheet explains these recommendations and what they might mean for you.

What is BRCA?

BRCA stands for “BREast CANcer susceptibility gene.” There are two main BRCA genes — *BRCA1* and *BRCA2*. (Genes carry all the biological information that is passed from parent to child.) The BRCA genes help a cell’s genetic material (DNA) function normally. However, mutations (harmful changes) in these genes can sometimes occur. When this happens, the cell’s DNA does not function correctly, and a woman’s risk of developing breast, ovarian, and some other cancers can be greatly increased.

Facts About Cancer and BRCA-related Mutations

Despite progress in screening and treatment, breast and ovarian cancer remain leading causes of cancer deaths in America. About 3 percent of American women die from breast cancer, and about 1 percent die from ovarian cancer.

Having a family history of breast or ovarian cancer may increase a woman’s risk of having cancer herself. In recent years, scientists have learned more about why family history may increase cancer risk. They discovered that certain mutations in the *BRCA1* and *BRCA2* genes in cells can greatly increase the risk for developing breast and ovarian cancer. These mutations can also increase the risk of other cancers, including those of the fallopian tube and peritoneum.

A family history of cancer may suggest that a woman is more likely to have a BRCA mutation. Of the women who do have a family history, however, very few have these gene changes.

Assessing Genetic Risk for Cancer and Reducing Risk for BRCA-related Cancer

Identifying women who have *BRCA1* and *BRCA2* mutations can help them and their health care professionals decide whether to take steps that can reduce their chances of developing cancer.

Assessing and testing for genetic risk is a process that involves several steps:

- Primary care professionals talk with all women about their family history of cancer. Depending on a woman's family history, the doctor or nurse may then use a special questionnaire to gather information, including details about the relations of family members, the types of cancers, and how old family members were when they developed cancer. These questions can help determine whether or not in-depth genetic counseling would be useful.
- If genetic counseling is needed, a woman will talk with a health care professional who is trained to do this kind of counseling. The counselor will conduct a detailed assessment of risk for BRCA mutations. The counselor also will explain about BRCA testing and the pros and cons of getting the test, and help the woman make the decision of whether or not to get tested.
- Based on the results of genetic counseling, the woman may choose to have a genetic test. The test involves examining her cells for changes in *BRCA1* and *BRCA2* genes. Genetic tests for BRCA mutations work best in women at high risk for having the mutation.
- Because BRCA test results are complex, it is critical that the woman discuss her test results with a trained counselor, who will explain what they mean and can help in decisions about possible next steps for treatment.

Based on the results of the BRCA test, the woman may choose a treatment that may reduce her chances of developing cancer. These treatments include frequent cancer screening, risk-reducing medications, or surgery to remove her breasts or ovaries.

Potential Benefits and Harms of Using Medications to Reduce Breast Cancer Risk

The Task Force reviewed recent studies on the benefits and harms of risk assessment, genetic counseling, genetic testing, and treatments aimed at reducing the risk for BRCA-related cancers.

*For Women Who **DO NOT** Have a Family History That is Associated With an Increased Risk for BRCA1 or BRCA2 Mutations*

Most women — more than 90 percent — do not have a family history that puts them at increased risk for having a BRCA mutation. As a result, genetic counseling, BRCA testing, and risk-reducing treatment have few or no benefits for most women.

BRCA testing in women who are not at risk has the potential for significant harms. Test results are often unclear and women have to live with uncertainty about whether they will or will not develop cancer. Unclear tests lead many women to take powerful medicines or have major surgery that they may not need.

*For Women Who **DO** Have a Family History That is Associated With an Increased Risk for BRCA1 or BRCA2 Mutations*

For these women, risk assessment, genetic counseling and BRCA testing, and risk-reducing treatment can have moderate benefits. The risk assessment questions can accurately predict which women could benefit from genetic counseling. Genetic counseling generally helps a woman understand what it might mean to her to have a gene mutation that may increase her chances of developing cancer. It also can help her make an informed decision about whether to have genetic testing.

Fewer than 10 percent of women have a family history that may put them at increased risk. Of the women with a family history who get genetic counseling:

- Many learn that they are actually not at increased risk or that they are not good candidates for the genetic test.
- Others, after learning about the benefits and risks, decide not to have the genetic test.
- Some learn they are good candidates and decide to get the genetic test; of these, very few find definitively that they have genes that may increase their chance of developing cancer.

The Task Force found limited evidence on whether interventions such as intensive screening and medications help reduce the risk of cancer for women who have a BRCA mutation. Some studies suggest that risk-reducing surgery may be effective.

The potential harms of risk assessment and genetic testing for women who are at risk are small to moderate. Tests results may not be clear, and treatment may have harms such as pain, side effects from medicines, or complications from surgery.

The Final Recommendations on Risk Assessment, Genetic Counseling, and Genetic Testing for BRCA-related Cancer in Women: What Do They Mean?

Here are the Task Force's final recommendations on risk assessment, genetic counseling, and genetic testing for BRCA-related cancer. Recommendations have letter grades. The grades are based on the quality and strength of the evidence about the potential benefits and harms of screening for this purpose. They also are based on the size of the potential benefits and harms. Task Force recommendation grades are explained in the box at the end of this fact sheet.

When the Task Force recommends counseling and genetic screening (**Grade B**), it is because they have more potential benefits than potential harms. When the Task Force recommends against counseling and genetic screening (**Grade D**), it is because they have more potential harms than potential benefits. The Notes explain key ideas.

Visit the Task Force Web site to read the full **final recommendation statement**. The statement explains the evidence the Task Force reviewed and how it decided on the grade. An **evidence report** provides more detail about the studies the Task Force reviewed.

1 The Task Force recommends that primary care providers screen women who have family members with breast, ovarian, **tubal**, or **peritoneal** cancer with one of several **screening tools** designed to identify a family history that may be associated with an increased risk for potential harmful **mutations** in breast cancer **susceptibility** genes (*BRCA1* and *BRCA2*). Women who screen positive should receive **genetic counseling** and, if indicated after counseling, **BRCA testing**. **Grade B**

2 The Task Force recommends against **routine** genetic counseling or BRCA testing of women whose family history is not associated with an increased risk for potentially harmful mutations in the *BRCA1* or *BRCA2* genes. **Grade D**

Notes

1 tubal

Cancer of the fallopian tubes. These two tubes connect a woman's ovaries to her uterus.

peritoneal

Cancer that develops in the peritoneum, the thin layer of tissue that lines the abdomen.

screening tools

Questionnaires.

mutations

Permanent changes to a cell's genetic information (DNA).

Susceptibility

Capable of being affected by something.

genetic counseling

Talking to a health care professional who is trained in genetic counseling. Counseling includes assessing in detail a person's risk for cancer, explaining genetic tests and possible results, and helping a person decide whether to get tested. Counseling also takes place after testing, to review and discuss results and what they may mean.

BRCA testing

Examining cells for changing to the *BRCA1* and *BRCA2* genes.

2 routine

Part of regular ongoing care.

Should You Get Genetic Counseling or Testing for BRCA-related Cancer Risk?

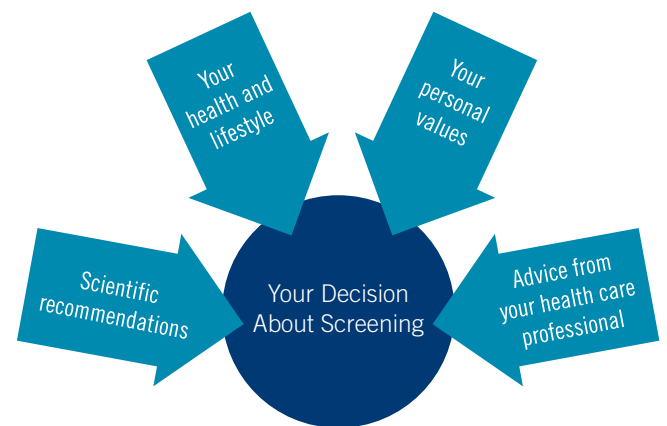
Getting the best health care means making smart decisions about what screening tests, counseling services, and preventive medications to get and when to get them. Many people don't get the tests, counseling, or medications they need. Others get tests, counseling, or medications they don't need or that may be harmful to them.

Task Force recommendations can help you learn about screening tests, counseling services, and preventive medications. These services can keep you healthy and prevent disease. The Task Force recommendations do not cover diagnosis (tests to find out why you are sick) or treatment of disease.

Task Force recommendations also apply to some groups of people, but not others. For example, this recommendation does not apply to women who are being treated for a BRCA-related cancer. It also doesn't apply to men.

Making a decision about genetic counseling and testing for BRCA-related cancer risk

Consider your own health and lifestyle. Think about your personal beliefs and preferences for health care. Talk with your health care professional about your family history of BRCA-related cancer and any other risk factors for cancer. Be comfortable that all your questions have been answered. And consider scientific recommendations, like this one from the Task Force. Use this information to become fully informed and to decide whether counseling and genetic testing are right for you.



What is the U.S. Preventive Services Task Force?






The Task Force is an independent group of national experts in prevention and evidence-based medicine. The Task Force works to improve the health of all Americans by making evidence-based recommendations about clinical preventive services such as screenings, counseling services, or preventive medicines. The recommendations apply to people with no signs or symptoms of the disease being discussed.

To develop a recommendation statement, Task Force members consider the best available science and research on a topic. For each topic, the Task Force posts draft documents for public comment, including a draft recommendation statement. All comments are reviewed and considered in developing the **final recommendation statement**. To learn more, visit the [Task Force Web site](#).

USPSTF Recommendation Grades

Grade	Definition
A	Recommended.
B	Recommended.
C	Recommendation depends on the patient's situation.
D	Not recommended.
I statement	There is not enough evidence to make a recommendation.

Click Here to Learn More About Breast Cancer and Strategies to Reduce Risk

-  [Talk with a Doctor if Breast or Ovarian Cancer Runs in Your Family](#) (healthfinder.gov)
-  [Genetic Testing for Breast and Ovarian Cancer: Questions for the Doctor](#) (healthfinder.gov)
-  [BRCA1 and BRCA2: Cancer Risk and Genetic Testing](#) (National Cancer Institute)
-  [What You Need to Know About Breast Cancer™](#) (National Cancer Institute)
-  [Ovarian Epithelial Cancer Treatment \(PDQ®\)](#) (National Cancer Institute)