

This fact sheet explains the U.S. Preventive Services Task Force's (Task Force) draft recommendation statement on risk assessment, genetic counseling, and genetic testing for BRCA-related cancer. It also tells you how you can send comments about the draft recommendation to the Task Force. Comments may be submitted from February 19, 2019 to March 18, 2019. The Task Force welcomes your comments.

Risk Assessment, Genetic Counseling, and Genetic Testing for BRCA-Related Cancer

The Task Force issued a **draft recommendation statement** on *Risk Assessment, Genetic Counseling, and Genetic Testing for BRCA-Related Cancer*.

The Task Force found that most women should not be screened, counseled, or tested for *BRCA1* or *BRCA2* mutations, unless they have a personal or family history of certain types of cancers — or have ethnicity or ancestry

associated with these mutations. Women who have a personal or family history should be screened to see if they are at increased risk for *BRCA1* and *BRCA2* genetic mutations. Women who are at increased risk should receive genetic counseling and, if indicated after counseling, offered BRCA testing.

What is BRCA?

BRCA stands for “BReast CAncer susceptibility gene.” There are two main BRCA genes — *BRCA1* and *BRCA2*. These genes help our bodies repair damaged DNA and help lower women’s likelihood of getting certain cancers. However, harmful mutations (changes) in these genes can sometimes occur. When this happens, a woman’s risk of developing breast, ovarian, and some other cancers can increase. One important step in preventing these cancers is helping women understand their risk.

Facts about BRCA Genes and BRCA-Related Cancer

Genes carry all the biological information that is passed from parent to child. BRCA genes, including *BRCA1* and *BRCA2*, are genes known to be related to cancer risk.

BRCA1 and *BRCA2* genes produce proteins that help our bodies repair damaged DNA, and as a result, help lower a woman’s chance of getting certain cancers. Cancers related to the BRCA genes include breast, ovarian, peritoneal (the thin layer of tissue that lines the abdomen), fallopian tube (the two tubes that connect a woman’s ovaries to her uterus), and pancreatic cancer.

When either of the BRCA genes is mutated (changed), it cannot repair damaged DNA and prevent cancer.

Fortunately, these mutations are very rare. Less than 1% of all women in the U.S. has a BRCA mutation. Some women are more likely to have a mutation, such as:

- Women who have had a BRCA-related cancer before age 40
- Ashkenazi Jewish women
- Women with a family history of breast or ovarian cancer

Mutations in the *BRCA1* and *BRCA2* genes are just one of many risk factors for these cancers, and a mutation does not mean the woman will get cancer.

Facts about Risk Assessment, Genetic Counseling, and Genetic Testing for BRCA-Related Cancer

Determining if a woman is at risk for BRCA-related cancer involves three steps:

- 1. Risk Assessment and Genetic Screening:** During a standard primary care office visit, clinicians ask patients whether or not they have a personal or family history of certain types of cancer or an ethnicity or ancestry associated with BRCA mutations. If she does, the clinician will use a screening tool to gather additional information about a woman's personal and family history and ethnicity and ancestry. For example, the clinician may ask about which family members had cancer, what specific types of cancers, or if she has Ashkenazi Jewish heritage. These questions can help determine whether or not in-depth genetic counseling is needed.
- 2. Genetic Counseling:** If genetic counseling is needed, a woman will talk with a professional who is trained to do this kind of counseling. The counselor will conduct a more detailed review of the patient's medical and family history to determine if genetic testing is appropriate. The counselor will also discuss the benefits and harms of genetic testing, and what to do if the test results show a BRCA mutation that is associated with an increased risk of cancer.
- 3. Genetic Testing:** Based on the results of genetic counseling, the counselor may recommend genetic testing, which is normally done by collecting a blood or saliva sample. Testing is complex and cannot definitively tell women if she has a potentially harmful mutation that will lead to cancer. That is why testing should be done by a trained genetic counselor or medical geneticist who can help women interpret their test results. Testing should begin with a relative who has had a BRCA-related cancer, including male relatives, before testing individuals without cancer.

Potential Benefits and Harms of Risk Assessment, Genetic Counseling, and Genetic Testing for BRCA-Related Cancer

The Task Force looked at evidence on the potential benefits and harms of risk assessment, genetic counseling, and genetic testing for BRCA-related cancer.

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

Most women do not have a personal or family history, ethnicity, or ancestry that puts them at increased risk for having a BRCA mutation. Screening, counseling, and testing have few or no benefits for these women, but there are real harms associated with these services. These harms can vary but may include psychologic harms such as increased worry before and after testing. In addition, treatment often involves risk-reducing medications, which can lead to unnecessary side effects, or surgery. Based on the evidence, the benefits do not outweigh the harms for these women.

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

For women who have a personal or family history, ethnicity, or ancestry associated with an increased risk for carrying a BRCA mutation, the Task Force found that there are benefits to screening, counseling, and testing for BRCA mutations. Benefits can include better understanding of breast cancer risk and reduction of BRCA-related cancers. While there are also the potential harms, such as increased worry associated with testing, as well as those associated with any needed treatment, the benefits outweigh the harms for these women.

The Draft Recommendations on Risk Assessment, Genetic Counseling, and Genetic Testing for BRCA-Related Cancer: What Do They Mean?

Here are the Task Force's draft recommendations on risk assessment, genetic counseling, and genetic testing for BRCA-related cancer. They are based on the quality and strength of the evidence about the potential benefits and harms of these services. They are also 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 issues a **B Grade**, it recommends a preventive service because it has more potential benefits than harms.

When the Task Force issues a **D Grade**, it recommends against a preventive service because the harms outweigh the benefits.

Before you send comments to the Task Force, you may want to read the **draft recommendation statement**. The recommendation statement explains the evidence the Task Force reviewed and how it decided on the grade. An **evidence document** provides more detail about the scientific studies the Task Force reviewed.

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.

ethnicity or ancestry
There are certain backgrounds that are associated with BRCA mutations, including Ashkenazi Jewish women.

BRCA
Breast CAncer susceptibility gene

mutations
Permanent changes to a cell's genetic information (DNA), which affects the cell's ability to do its job. For *BRCA1* and *BRCA2* genes this means that it cannot do its main job of repairing damaged DNA and preventing the development of cancers.

susceptibility genes
Being more likely to be affected by a disease because of your genetic makeup.

2 **routine**
Part of regular care. These services should not be done for women not at increased risk for mutations of the *BRCA1* or *BRCA2* genes.

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

2 The USPSTF recommends against **routine** genetic counseling or BRCA testing for women whose family history or ethnicity/ancestry is not associated with an increased risk for potentially harmful mutations in the *BRCA1* or *BRCA2* genes. **(D Grade)**





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

The Task Force is an independent, volunteer 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, and 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 BRCA

-  **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)
-  **BRCA Mutations: Cancer Risk and Genetic Testing**
(National Cancer Institute)
-  **Hereditary Breast Cancer and BRCA Genes**
(Centers for Disease Control and Prevention)

[Click Here](#) to Comment on the Draft Recommendation



The Task Force welcomes comments on this draft recommendation.



Comments must be received between February 19, 2019 and March 18, 2019.



All comments will be considered for use in writing final recommendations.