

## Genetic Risk Assessment and *BRCA* Mutation Testing for Breast and Ovarian Cancer Susceptibility: U.S. Preventive Services Task Force Recommendations

*Summaries for Patients* are a service provided by *Annals* to help patients better understand the complicated and often mystifying language of modern medicine.

The full reports are titled “Genetic Risk Assessment and *BRCA* Mutation Testing for Breast and Ovarian Cancer Susceptibility: Recommendation Statement” and “Genetic Risk Assessment and *BRCA* Mutation Testing for Breast and Ovarian Cancer Susceptibility: Systematic Evidence Review for the U.S. Preventive Services Task Force.” They are in the 6 September 2005 issue of *Annals of Internal Medicine* (volume 143, pages 355-361 and 362-379). The first report was written by the U.S. Preventive Services Task Force; the second report was written by H.D. Nelson, L. Hoyt Huffman, R. Fu, and E.L. Harris.

### Who developed these guidelines?

The U.S. Preventive Services Task Force (USPSTF) is a group of health experts that reviews published research and makes recommendations about preventive health care.

### What is the problem and what is known about it so far?

Many women with breast cancer have first-degree (mother, sister, daughter) or second-degree (aunt) relatives with breast cancer. Mutations on 2 genes have been shown to increase the risk for breast and ovarian cancer: breast cancer susceptibility genes 1 and 2 (*BRCA1* and *BRCA2*). These mutations are more common in certain ethnic groups, such as Ashkenazi Jewish individuals. However, not all women with a family history of breast or ovarian cancer have one of these mutations and not all women with one of these mutations will develop cancer. Genetic testing could identify women who should consider intense cancer screening or procedures to reduce cancer risk (medication or surgical removal of breasts, ovaries, or both). However, since not all women who have a *BRCA* mutation develop cancer, identification of mutations may also needlessly expose women to anxiety, insurance problems, or unnecessary procedures.

### How did the USPSTF develop these recommendations?

The USPSTF reviewed published research to evaluate the benefits and harms of genetic assessment and *BRCA* mutation testing for breast and ovarian cancer susceptibility.

### What did the authors find?

Little is known about *BRCA* mutations in the general population. Most studies of *BRCA* mutations have been done in women with existing cancer or strong family histories of cancer. Studies do not definitively prove that intensive screening improves cancer outcomes in women with *BRCA* mutations. Using medications to prevent cancer in women at high risk does reduce cancer, but these medications also cause side effects. Surgical removal of the breasts and ovaries does reduce the development of cancer in women with *BRCA1* and *BRCA2* mutations. However, surgery always carries the risk for complications and these surgeries can decrease quality of life for some women.

### What does the USPSTF suggest that patients and doctors do?

Doctors and patients should be aware of the family history patterns that are associated with an increased risk for *BRCA* mutations. In women who are not of Ashkenazi (Eastern European) Jewish heritage, these include 2 first-degree relatives with breast cancer, at least 1 of whom received a diagnosis at age 50 years or younger; 3 or more first- or second-degree relatives with breast cancer diagnosed at any age; a first-degree relative with bilateral breast cancer diagnosed at any age; a combination of 2 or more first- or second-degree relatives with ovarian cancer diagnosed at any age; and a male relative with breast cancer. For women of Ashkenazi (Eastern European) Jewish heritage, these include any first-degree relative with breast or ovarian cancer at any age or at least 2 second-degree relatives on the same side of the family with breast or ovarian cancer at any age.

The USPSTF recommends against referral for genetic evaluation if a woman's family history does not suggest an increased risk for *BRCA* mutations. The USPSTF recommends referral of women whose family history is associated with an increased risk for harmful mutations in *BRCA1* or *BRCA2* to suitably trained health care providers.

### What are the cautions related to these recommendations?

These recommendations do not apply to women with a personal history of breast or ovarian cancer or with a family member who has a known *BRCA* mutation.

Summaries for Patients are presented for informational purposes only. These summaries are not a substitute for advice from your own medical provider. If you have questions about this material, or need medical advice about your own health or situation, please contact your physician. The summaries may be reproduced for not-for-profit educational purposes only. Any other uses must be approved by the American College of Physicians.