

Putting Prevention into Practice

An Evidence-Based Approach

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

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Case Study

A 31-year-old woman presents to your office for a well-woman examination. She is sexually active in a monogamous relationship and has never been pregnant. She tells you that her mother was diagnosed with tubal cancer at age 40 and that her sister, who is 42 years of age, was recently diagnosed with breast cancer. On further questioning, she reveals that her maternal aunt was diagnosed with breast cancer at 45 years of age. Physical examination shows no abnormal breast masses or lumps.

Case Study Questions

1. Based on the U.S. Preventive Services Task Force (USPSTF) recommendation statement, which one of the following is the most appropriate next step for this patient?
 - A. Refer her to a geneticist for genetic testing.
 - B. Assess her risk for a potentially harmful *BRCA1/2* mutation using an appropriate brief familial risk assessment tool.
 - C. Refer her for breast cancer screening using mammography, breast ultrasonography, or both.
 - D. Recommend monthly breast self-examinations.
 - E. Recommend yearly clinical breast examinations.
2. According to the USPSTF recommendation, which of the following risk assessment instruments accurately estimate the likelihood of carrying a harmful *BRCA1/2* mutation?
 - A. Ontario Family History Assessment Tool.
 - B. 7-Question Family History Screening Tool.
 - C. International Breast Cancer Intervention Study instrument (Tyrer-Cuzick).
 - D. National Cancer Institute Breast Cancer Risk Assessment Tool (Gail Model).
3. You determine that the patient has an elevated risk of a potentially harmful *BRCA1/2* mutation. According to the USPSTF, which one of the following is the most appropriate next step?
 - A. Order genetic testing to screen for a *BRCA1/2* mutation.
 - B. Order genetic testing for the patient's mother to screen for a *BRCA1/2* mutation.
 - C. Provide or refer the patient to genetic counseling and, if indicated, genetic testing.
 - D. Offer to prescribe medications to reduce the patient's risk of breast cancer.
 - E. Refer the patient for twice-yearly mammography starting at 40 years of age.

Answers appear on the following page.

See related U.S. Preventive Services Task Force Recommendation Statement on page 233.

This PPIP quiz is based on the recommendations of the USPSTF. More information is available in the USPSTF Recommendation Statement and supporting documents on the USPSTF website (<https://www.uspreventiveservicestaskforce.org>). The practice recommendations in this activity are available at <https://www.uspreventiveservicestaskforce.org/Page/Document/UpdateSummaryFinal/brca-related-cancer-risk-assessment-genetic-counseling-and-genetic-testing1>.

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A collection of Putting Prevention into Practice published in *AFP* is available at <https://www.aafp.org/afp/ppip>.

CME This clinical content conforms to AAFP criteria for continuing medical education (CME). See CME Quiz on page 199.

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Answers

1. The correct answer is B. The USPSTF recommends that primary care clinicians assess women with a personal or family history of breast, ovarian, tubal, or peritoneal cancer or who have an ancestry associated with breast cancer susceptibility 1 and 2 (*BRCA1/2*) gene mutations with an appropriate brief familial risk assessment tool. Women with a positive result on the risk assessment tool should then receive genetic counseling and, if indicated after counseling, genetic testing (B recommendation).¹ In this case, the patient has several family members with a history of cancers related to *BRCA1/2* mutations, and her risk for harmful mutations should be assessed first. Personal and family history of suspicious cancers should also be reassessed periodically.

2. The correct answers are A, B, and C. Validated tools evaluated by the USPSTF include the Ontario Family History Assessment Tool, Manchester Scoring System, Referral Screening Tool, Pedigree Assessment Tool, 7-Question Family History Screening Tool, International Breast Cancer Intervention Study instrument (Tyrer-Cuzick), and brief versions of BRCAPRO.² They can be used to guide referrals to genetic counseling for more definitive risk assessment.¹ General breast cancer risk assessment models, such as the National Cancer Institute Breast Cancer Risk Assessment Tool (Gail Model), are not designed to identify *BRCA*-related cancer risk and should not be used for this purpose.

3. The correct answer is C. The patient should receive genetic counseling. Genetic counseling about *BRCA1/2* mutation testing should be performed by trained health professionals, including suitably trained primary care clinicians. The process of genetic counseling includes detailed kindred analysis and risk assessment for potentially harmful *BRCA1/2* mutations. It also includes identification of candidates for testing, patient education, discussion of the benefits and harms of genetic testing, interpretation of results after testing, and discussion of management options. Patients should not be tested for harmful mutations before receiving appropriate counseling. Interventions such as risk-reducing medications or intensive interventions may be considered, although only after genetic counseling and genetic testing.

The views expressed in this work are those of the authors and do not reflect the official policy or position of Case Western Reserve University/University Hospitals Cleveland Medical Center, the U.S. Department of Health and Human Services, or the U.S. government.

References

1. Owens DK, Davidson KW, Krist AH, et al. Risk assessment, genetic counseling, and genetic testing for *BRCA*-related cancer: US Preventive Services Task Force recommendation statement [published correction appears in *JAMA*. 2019;322(18):1830]. *JAMA*. 2019;322(7):652-665.
2. Nelson HD, Pappas M, Cantor A, et al. Risk assessment, genetic counseling, and genetic testing for *BRCA*-related cancer in women: updated evidence report and systematic review for the US Preventive Services Task Force. *JAMA*. 2019;322(7):666-685. ■