

Editorials

Integrating Breast Cancer Risk Management into Primary Care

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See related U.S. Preventive Services Task Force Recommendation Statement at <https://www.aafp.org/afp/2020/0315/od1.html> and related Putting Prevention into Practice on page 373.

In 2019, the U.S. Preventive Services Task Force (USPSTF) updated two recommendation statements related to breast cancer: the *BRCA*-related cancer risk assessment, including evaluating for genetic counseling and testing,¹ and the use of preventive medication to reduce breast cancer risk.² Because approximately 10% of breast cancers are attributable to a genetic mutation (also called a pathogenic variant) and at least 11 genes are now known to increase the risk of breast cancer,^{3,4} it is more important than ever that primary care physicians have the ability and resources to identify women eligible for genetic counseling and testing.

Who Should Be Screened for Genetic Mutations?

Women with a family history of breast, ovarian, tubal, or peritoneal cancer and previously treated survivors of any of those cancers should be assessed for genetic risk. More than 16 million

individuals are living with a history of cancer in the United States,⁵ and more than 70% of cancer survivors receive health care from their primary care physician.⁶ Individuals with a mutation may be eligible for high-risk breast cancer screening with annual mammography and breast magnetic resonance imaging, as well as risk-reducing measures such as surgery or chemoprevention.

Useful Screening Tools

Family physicians should collect, review, and update patients' personal and family history annually. Validated screening tools (*Table 1*⁷⁻¹¹) are available for use in the clinic for identification of patients eligible for genetic risk assessment. The 7-Question Family History Screening tool is a patient-completed questionnaire; a single positive response should trigger recommendations for genetic risk assessment.¹¹ The Pedigree Assessment Tool is also brief and easy to use, but it requires scoring.¹⁰ This tool would be best utilized by the clinician team to screen patients based on documented history. Each practice should identify a genetic expert for counseling and testing and should know where to refer patients after a pathogenic mutation has been identified.

Risk Assessment and Chemoprevention

Beyond genetic risk assessment, all women without a personal history of breast cancer should be assessed for individualized breast cancer risk.

TABLE 1

Screening Tools for Breast and Ovarian Cancer Genetic Risk Assessment

Tool	Website
Breast Cancer Genetics Referral Screening Tool ⁷	https://www.breastcancergenescreeen.org/
Manchester scoring system ⁸	https://www.researchgate.net/figure/Manchester-scoring-system_tbl1_7749080
Ontario Family History Assessment Tool ⁹	https://www.timeofcare.com/ontario-family-history-assessment-tool/
Pedigree Assessment Tool ¹⁰	https://www.timeofcare.com/pedigree-assessment-tool/
7-Question Family History Screening tool ¹¹	https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2739222/table/T1/

Information from references 7-11.

Breast cancer risk assessments have not been shown to increase worry and anxiety for women, and they improve understanding of breast cancer risk.^{12,13} Use of the recently updated USPSTF recommendation requires physicians to quantify an individual's risk of breast cancer and to identify women who would most likely benefit from chemoprevention.² Most clinical trials assessing the effectiveness of risk-reducing medications (e.g., tamoxifen, raloxifene [Evista], aromatase inhibitors) used the Breast Cancer Risk Assessment Tool, also known as the Gail model, to define a woman as high risk with a threshold five-year breast cancer risk of greater than 1.66%.¹⁴

The updated USPSTF recommendations suggest that physicians offer risk-reducing medications to women at increased risk of breast cancer.² These antihormonal therapies have been shown to reduce the risk of breast cancer by 40% to 60% but have not demonstrated a survival benefit.¹⁵⁻¹⁸ The Gail model can be completed in less than 60 seconds, but the shared decision-making process regarding chemoprevention is time-consuming. To help women make individualized decisions, physicians should discuss not only breast cancer risk, but also the adverse effects of medications, which include vasomotor symptoms, vaginal symptoms, and bone effects. Increased risk of uterine cancer occurs only with use of tamoxifen.

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References

1. U.S. Preventive Services Task Force. *BRCA*-related cancer: risk assessment, genetic counseling, and genetic testing. August 2019. Accessed January 6, 2020. <https://www.uspreventiveservicestaskforce.org/Page/Document/UpdateSummaryFinal/brca-related-cancer-risk-assessment-genetic-counseling-and-genetic-testing1>
2. U.S. Preventive Services Task Force. Breast cancer: medication use to reduce risk. September 2019. Accessed January 6, 2020. <https://www.uspreventiveservices.org/Page/Document/UpdateSummaryFinal/breast-cancer-medications-for-risk-reduction1>
3. Beitsch PD, Whitworth PW, Hughes K, et al. Underdiagnosis of hereditary breast cancer: are genetic testing guidelines a tool or an obstacle? *J Clin Oncol*. 2019;37(6):453-460.
4. National Comprehensive Cancer Network. Genetic/familial high risk assessment: breast, ovarian, and pancreatic; version 1.2020. Accessed January 14, 2020. https://www.nccn.org/professionals/physician_gls/default.aspx#genetics_screening

5. American Cancer Society. Cancer treatment and survivorship: facts and figures 2019-2021. Accessed January 6, 2020. [https://www.cancer.org/content/dam/cancer-org/research/cancer-facts-and-statistics/cancer-treatment-and-survivorship-facts-and-figures-2019-2021.pdf](https://www.cancer.org/content/dam/cancer-org/research/cancer-facts-and-statistics/cancer-treatment-and-survivorship-facts-and-figures/cancer-treatment-and-survivorship-facts-and-figures-2019-2021.pdf)
6. Kansas Department of Health and Environment. Cancer survivorship among Kansas adults: 2016 Kansas behavioral risk factor surveillance system. March 2019. Accessed January 6, 2020. http://www.kdheks.gov/cancer/download/Kansas_Survivorship_Among_Kansas_Adults_March_2019.pdf
7. Gilpin CA, Carson N, Hunter AG. A preliminary validation of a family history assessment form to select women at risk for breast or ovarian cancer for referral to a genetics center. *Clin Genet*. 2000;58(4):299-308.
8. Oros KK, Ghadirian P, Maugard CM, et al. Application of *BRCA1* and *BRCA2* mutation carrier prediction models in breast and/or ovarian cancer families of French Canadian descent. *Clin Genet*. 2006;70(4):320-329.
9. Bellcross CA, Lemke AA, Pape LS, et al. Evaluation of a breast/ovarian cancer genetics referral screening tool in a mammography population. *Genet Med*. 2009;11(11):783-789.
10. Teller P, Hoskins KF, Zwaagstra A, et al. Validation of the pedigree assessment tool (PAT) in families with *BRCA1* and *BRCA2* mutations. *Ann Surg Oncol*. 2010;17(1):240-246.
11. Ashton-Prolla P, Giacomazzi J, Schmidt AV, et al. Development and validation of a simple questionnaire for the identification of hereditary breast cancer in primary care. *BMC Cancer*. 2009;9:283.
12. Livaudais-Toman J, Karliner LS, Tice JA, et al. Impact of a primary care based intervention on breast cancer knowledge, risk perception and concern: a randomized, controlled trial. *Breast*. 2015;24(6):758-766.
13. Helmes AW, Culver JO, Bowen DJ. Results of a randomized study of telephone versus in-person breast cancer risk counseling. *Patient Educ Couns*. 2006;64(1-3):96-103.
14. National Cancer Institute. The Breast Cancer Risk Assessment Tool. Accessed January 6, 2020. <https://bcrisktool.cancer.gov/index.html>
15. Cuzick J, Sestak I, Cawthorn S, et al.; IBIS-I Investigators. Tamoxifen for prevention of breast cancer: extended long-term follow-up of the IBIS-I breast cancer prevention trial. *Lancet Oncol*. 2015;16(1):67-75.
16. Cuzick J, Sestak I, Forbes JF, et al.; IBIS-II Investigators. Anastrozole for prevention of breast cancer in high-risk postmenopausal women (IBIS-II): an international, double-blind, randomised placebo-controlled trial [published correction appears in *Lancet*. 2014;383(9922):1040]. *Lancet*. 2014;383(9922):1041-1048.
17. Fisher B, Costantino JP, Wickerham DL, et al. Tamoxifen for the prevention of breast cancer: current status of the National Surgical Adjuvant Breast and Bowel Project P-1 study. *J Natl Cancer Inst*. 2005;97(22):1652-1662.
18. Vogel VG, Costantino JP, Wickerham DL, et al.; National Surgical Adjuvant Breast and Bowel Project (NSABP). Effects of tamoxifen vs raloxifene on the risk of developing invasive breast cancer and other disease outcomes: the NSABP Study of Tamoxifen and Raloxifene (STAR) P-2 trial [published corrections appear in *JAMA*. 2007;298(9):973 and *JAMA*. 2006;296(24):2926]. *JAMA*. 2006;295(23):2727-2741. ■