New BRCA recommendations: What primary care doctors must know

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Kevin B. O'Reilly
News Editor

The U.S. Preventive Services Task Force (USPSTF) has updated its recommendations on screening for the \textit{BRCA1} and \textit{BRCA2} genetic mutations that account for 15\% of ovarian cancer cases and between 5\% and 10\% of breast cancer cases. Here is what primary care physicians should know about the new recommendations.

The USPSTF’s recommendation statement, published in \textit{JAMA} along with the task force’s evidence report, represents a significant change to the group’s 2013 recommendations and broadens the pool of women for whom primary care physicians should conduct a risk assessment and potentially refer for genetic counseling and testing.

Previously, the USPSTF recommended risk assessment for women with a family history of breast cancer, ovarian cancer or both. Now that’s been broadened to include women who previously had ovarian or breast cancer but are now cancer-free, as well as women with ancestry associated with \textit{BRCA1} or \textit{BRCA2} genetic mutations. In the latter group, there’s a well-established link between Ashkenazi Jewish ancestry and the mutations.

“The addition of women with prior breast and ovarian cancer is an important step forward,” wrote Susan Domchek, MD, and Mark Robson, MD, of the University of Pennsylvania Basser Center for BRCA, in a \textit{JAMA} editorial on the USPSTF statement.

Drs. Domchek and Robson noted that \textit{BRCA1} and \textit{BRCA2} “status is relevant for patients with newly diagnosed early stage breast cancer for surgical decision-making and can also be used to determine appropriate treatment of certain advanced cancers.” Identifying individuals “at risk of carrying a \textit{BRCA1/2} mutation can be lifesaving and should be a part of routine medical care,” they added.

Too few patients
The new grade-B USPSTF recommendations come amid evidence that most patients who could benefit from BRCA testing are not getting it. In their editorial, Drs. Domchek and Robson noted that less than 30% of patients with epithelial ovarian cancer are tested for BRCA mutations, even though about 15% of women who have the cancer also have a BRCA1 or BRCA2 pathogenic variant.

About one in 300 people in the general population has one of three BRCA1 or BRCA2 mutations, Drs. Domchek and Robson wrote. That compares to one in 40 people of Ashkenazi Jewish descent, of which there are between 5 million and 6 million in the U.S.

The task force did not endorse any particular risk-assessment tools. However, the USPSTF noted several reliable options: the Ontario Family History Assessment Tool, Manchester Scoring System, Referral Screening Tool, Pedigree Assessment Tool, 7-Question Family History Screening Tool, and the International Breast Cancer Intervention Study instrument.

If primary care physicians wind up referring patients for genetic counseling and testing, another complication arises, Drs. Domchek and Robson explained. Large genetic panels testing for 80 or more genes have largely taken hold, and it is difficult to find tests solely for the BRCA1 or BRCA2 variants of interest.

The clinical usefulness of these multigene panel tests has not been established, says the JAMA editorial, “and genetics education of primary care clinicians has not kept pace with either the influx of new information or the changes in the genetic testing marketplace.”

You can find more commentary on the USPSTF recommendation across the JAMA Network™ journals. The following editorials commenting on the task force statement were published:


JAMA also has published an open-access article that you can share with interested patients that is called “Should I Be Tested for BRCA Mutations?”


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