New Guidelines for Assessing BRCA1/2-Related Cancers

BY MARY BROPHY MARCUS

In late August, the U.S. Preventive Services Task Force (USPSTF) issued a new recommendation statement on risk assessment, genetic counseling, and genetic testing for BRCA1/2-related cancer in women.

Inherited mutations in BRCA1 and BRCA2 are linked to an increased risk of developing breast, ovarian, peritoneal, and fallopian tube cancers. According to the NIH, women who inherit one of the BRCA genes are at a much higher risk for cancer. For example, approximately 72 percent of women who inherit a harmful BRCA1 mutation and about 69 percent of women who inherit a harmful BRCA2 mutation develop breast cancer by the time they’re 80 years old. In the general population, the statistic is much lower, with about 12 percent of women developing breast cancer sometime during their lives.

The new USPSTF recommendations say primary care clinicians should assess women who have a personal or family history of breast, ovarian, tubal, or peritoneal cancer. Patients who have an ancestry associated with BRCA1/2 gene mutations should also be assessed for increased risk of the BRCA1 and BRCA2 genetic mutations. Women who come up positive on the risk assessment should then pursue genetic counseling and possibly genetic testing if counseling supports that step.

The USPSTF does not recommend routine risk assessment, genetic counseling, or genetic testing for women without the harmful BRCA1/2 gene mutations.

“I agree with the recommendations. Women with a family history of breast cancer should be referred for genetic counseling,” stated Anna Weiss, MD, Assistant Professor of Surgery at Harvard Medical School and breast surgical oncologist at Brigham and Women’s Hospital.

Weiss believes tasking primary care physicians to do risk assessments is key. “Although our internal medicine workforce is overworked and underappreciated, often they are still our first line of defense. We see patients who come from these kinds of referrals all the time,” she said.

Patients who know they have a family history of BRCA1/2-related breast cancer should also be proactive. “Our high-risk clinics are probably underutilized. If someone were to have a family history, screening is important,” Weiss added.

She noted that primary care physicians should also be looking at other risk factors for breast cancer, which the new recommendations don’t cover, including lifestyle factors such as obesity, smoking, alcohol intake, and diet. “All of these things carry with them a potential risk of breast cancer.”

Weiss noted that age plays a role, too. Women with a family history of BRCA1/2-related breast cancers are at a higher risk above their peers of developing a breast cancer between the ages of 30 and 50.

“Their risk is higher then, in those earlier years, compared to a person who does not have a genetic mutation, and that’s when it is most beneficial to detect these mutations. Not to be ageist, though. For patients who are older, it may be important to detect it not only for them, but for their progeny,” Weiss stated.

She also agrees with the second piece of the task force recommendation regarding genetic counseling; in the absence of a family history of breast cancer, “the chances of having a BRCA1/2 are pretty low.”

Alice Police, MD, Regional Director of Breast Care at Northwell Health Cancer Institute, shared her thoughts on the new recommendations. “I think it’s great. Primary care doctors do need to take a more active role in patients with these family histories. I think PCPs should pay attention to family history. They should be thinking about genetics. They should use tools to identify patients at high risk for breast and ovarian cancer,” she noted. But once identified, she believes patients should move on to specialists.

“Here at Northwell Health, we have very specific high-risk breast screening program that’s really beyond the scope of a primary care doctor. But we’d like the primary care doctors to get those patients over to us,” Police said.

A third expert weighed in on screening by primary care physicians. “I think PCPs are definitely qualified,” stated Banu Arun, MD, a breast medical oncologist and Co-Medical Director of the MD Anderson Cancer Genetics Program, noting there are multiple tools they can use.

Evaluated by the USPSTF, the following tools have been validated and can accurately estimate the likelihood of carrying a harmful BRCA1/2 gene (JAMA 2019;322(7):652-665):

- 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)

However, general breast cancer risk assessment models, such as the National Cancer Institute Breast Cancer Risk Assessment Tool, are not designed to identify BRCA-related cancer risk, the USPSTF noted.

Arun also stated that the recommendations don’t cover other genes related to breast cancer and hopes to see future screening guidelines developed for those.

Mary Brophy Marcus is a contributing writer.