Panel: No Routine Screening for BRCA Mutations

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Women should not routinely undergo genetic screening or counseling for breast and ovarian cancer risk based on gene mutations unless they are at high risk, according to a first-time recommendation on this topic by the U.S. Preventive Services Task Force.

The task force, an independent panel of experts in prevention and primary care, recommended that primary care physicians not routinely refer women for genetic counseling or DNA testing for *BRCA1* or *BRCA2* gene mutations because harms and costs outweigh the benefits (Ann. Intern. Med. 2005;143:355-61).

However, the task force did recommend that women with a family history of breast or ovarian cancer undergo genetic screening for gene mutations if they are at increased risk.

Women who are at increased risk include those of Ashkenazi Jewish descent with a first- or second-degree relative with breast or ovarian cancer. Women not of this descent are at risk if they have family history patterns of breast and ovarian cancer (multiple first- and second-degree relatives).

According to the task force, only 2% of the general population is said to have *BRCA* gene mutations based on family history.

The task force recommendation estimated that between 1 in 300 and 1 in 500 women have the *BRCA* gene mutations in the general population, but not everyone with the mutations will develop breast or ovarian cancer.

Of women aged 70 years and younger with *BRCA* gene mutations, 35%-84% will develop breast cancer and 10%-50% will develop ovarian cancer, the task force noted.

In women who have been found to have a *BRCA1* or *BRCA2* gene mutation, MRI has proved to be more effective than other screening methods to detect breast cancer.

Options to reduce breast and ovarian cancer risk in women who are found to have *BRCA* gene mutations include mastectomy or oophorectomy, intensive screening, preventive chemotherapy, or a combination of preventive measures, according to the task force, adding that the benefits of taking these steps have not been confirmed.

The panel said that women with a family history of breast or ovarian cancer should undergo genetic screening for gene mutations if they are at increased risk.

The task force recommended that physicians and patients work together to decide what if any preventive measures to pursue in patients with the gene mutations.

"A woman who gains an understanding of the risk she faces may feel less anxious and have a sense of better control of her future," said Ned Calonge, M.D., the chair of the task force.

Tools are available to assess *BRCA1* and *2* gene mutation risk, but their effectiveness in the primary care setting has not yet been determined, according to the task force.

Further studies on this topic should include "the effectiveness of risk stratification and genetic counseling when delivered in different settings and by different types of providers, appropriate training for counselors, use of system supports, and patient acceptance of educational strategies," the task force wrote in its recommendation.

Breast cancer mortality should be assessed in future studies based on women who undergo an MRI. In addition, *BRCA* screening tools should be studied and validated for use in primary care to help physicians make appropriate referrals for genetic counseling, according to the task force.

In 2000, the American College of Obstetricians and Gynecologists recommended offering *BRCA* testing to people who have multiple family members with breast or ovarian cancer and have been found to have the gene mutation. ■

The task force recommendations are available online at http://www.ahrq.gov/clinic/uspstf/uspsbrca.htm.

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