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Caring for Young Women Who Are at High Risk for EARLY-ONSET BREAST CANCER

A Summary of Screening, Counseling, and Testing Guidelines

Health care providers can play a critical role in helping to reduce the incidence of hereditary breast and ovarian cancer by identifying patients with elevated risk. The following guidelines can help identify young women who may have a higher risk for hereditary breast and ovarian cancer and detect early-onset breast cancer among these women. The recommendations are based on current peer-reviewed evidence for screening, counseling, and testing.

Who is at high risk for early-onset breast cancer?

According to the American Cancer Society (ACS), women who are at high risk for early-onset breast cancer include those who:

- Have a known *BRCA1* or *BRCA2* gene mutation.
- Have a first-degree relative* (parent, brother, sister, or child), second-degree relative (aunts, uncles, nieces, or grandparents), or third-degree relative, which includes first cousins, with a *BRCA1* or *BRCA2* gene mutation.
- Have a lifetime risk of breast cancer of about 20% to 25% or greater, according to **risk assessment tools** **https://www.cdc.gov/cancer/breast/young_women/bringyourbrave/health_care_provider_education/risk_assessment_tools.htm** that are based mainly on family history.
- Had radiation therapy to the chest between the ages of 10 and 30 years.
- Have Li-Fraumeni syndrome, Cowden syndrome, or Bannayan-Riley-Ruvalcaba syndrome, or have first-degree relatives with one of these syndromes.

What are the current genetic counseling and testing guidelines for women at high risk for breast cancer?

The ACS recommends genetic testing for women at high risk, as defined above, to look for mutations in the *BRCA1* and *BRCA2* genes (or less commonly in other genes such as *PTEN* or

*Risk is higher with more than one affected first-degree relative and if the affected relative was diagnosed at a young age.

TP53). Although testing can be helpful in some situations, providers need to weigh the pros and cons with the patient. The ACS strongly recommends that women first talk to a genetic counselor, nurse, or doctor who is qualified to explain and interpret the results of these tests.

The U.S. Preventive Services Task Force (USPSTF) recommends that women who have one or more family members with a known potentially harmful mutation in the *BRCA1* or *BRCA2* genes should be offered genetic counseling and testing. Women with an identified increased risk should be referred to a genetic counselor, who can further evaluate the risk based on family history, discuss the pros and cons of testing, and arrange for *BRCA* testing if the patient is ready to proceed.

What are the current screening guidelines for women at high risk for breast cancer?

The National Comprehensive Cancer Network (NCCN) recommends screening for women with a *BRCA1* or *BRCA2* mutation or a first-degree relative who has a *BRCA1/2* mutation, even if the patient has not been tested for *BRCA1/2* mutations.

The NCCN recommends that women at high risk get a mammogram and breast MRI every year starting at age 25 to 40, depending on the type of gene mutation and/or youngest age of breast cancer in the family. The NCCN also suggests that women at high risk have clinical breast exams every 6 to 12 months beginning at age 25. These women should also consult with a health care provider to weigh the pros and cons of 3D mammography and learn how to identify changes in their breast.

The ACS recommends that women at high risk should get a mammogram and breast MRI every year. ACS suggests that women at high risk begin their screening at age 30 or an age recommended by their health care provider and continue for as long as they are in good health. Since there is limited evidence for the best age to start screening, ACS believes that this decision should be shared by the patient and her health care provider to ensure personal circumstances and preferences are taken into account.

What are the possible next steps after screening women at high risk for early-onset breast cancer?

The strategies to manage and reduce risk for women with a confirmed genetic susceptibility to breast and ovarian cancer include earlier, more frequent, or additional and intensive cancer screening modalities (such as breast MRI); risk-reducing surgeries (such as mastectomy or salpingo-oophorectomy), and medications (such as tamoxifen). Personal preference is an important factor in patients' decisions about risk-reducing strategies.

Additional Resources

American Cancer Society: Breast Cancer Early Detection and Diagnosis
http://bit.ly/ACSBreastCancerScreening

American Cancer Society Genetic Testing: Understanding Genetic Testing for Cancer (PDF)
http://bit.ly/ACSUnderstandingGeneticTesting

USPSTF Current Breast Cancer Screening Recommendations by Age Group
http://bit.ly/USPSTFBreastCancerScreening

USPSTF BRCA-Related Cancer: Risk Assessment, Genetic Counseling, and Genetic Testing
http://bit.ly/USPSTFBRCARelatedCancer

More Information
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