Current Breast Cancer Screening Guidelines Including for At-Risk Women

The American College of Radiology updated their guidelines for mammographic screenings recently. The ACR strongly supports early screening with particular detail to at-risk women. The ARC Commission on Breast Imaging evaluated numerous studies regarding effective methods for screening women at higher risk for breast cancer. The evaluation utilized the American College of Radiology’s Appropriateness Criteria® to help ensure that clinicians use the latest evidence-based methodology utilizing digital mammogram, digital breast tomosynthesis (DBT), molecular breast imaging (MBI), MRI and ultrasound.

The ACR and Society of Breast Imaging (SBI) endorse the premise that early detection reduces the death rate of women diagnosed with breast cancer.

Criteria for At-Risk Women

The ACR guidelines provide guide for clinicians to help identify at-risk women. Every female patient should be evaluated for their lifetime breast cancer risk, based on family history, breast density, genetic mutation status, ethnicity, childhood cancer status, and personal history of breast cancer.

Women with genetic mutations. The most widely recognized mutations for breast cancer are the BRCA1 or BRCA2 mutation. Women with the BRCA1 mutation carry a 50-to-85 percent risk of developing breast cancer in their lifetime. BRCA2 carriers have a 45 percent risk within their lifetime. Women who are of Ashkenazi Jewish descent are at a higher risk of having the BRCA genetic mutation but are also at higher risk for other
actionable mutations. Less common mutations include:

- ATM also called ataxiatelangiectasia genes
- CDH1 referred to as hereditary diffuse gastric cancer
- PALB2 which interacts with BRCA2
- PTEN, known as the Cowden and Bannayan-Riley Ruvalcaba syndromes
- STK11 known as Peutz-Jeghers syndrome
- TP53 and CHEK2, known as the Li-Fraumeni syndrome

Women with a strong family history. These women pose a special challenge to clinicians. Women who have first-and-second degree relatives diagnosed with breast cancer, particularly at an early age, are at increased risk. The Tyrer-Cuzick model appears to be the most accurate method to access a woman’s risk of developing breast cancer.

Women who received chest or mantle radiation therapy as a child or young woman. Typically, these women are childhood Hodgkin’s Lymphoma survivors. The increased risk for breast cancer begins about eight years after their radiation. A cumulative dose >10 Gy given before a women reaches her 30th birthday puts her at high risk for breast cancer.

Women with a personal history of breast cancer. Studies show that women who received breast-conserving therapy face an almost 20 percent risk of recurrence within ten years. A woman’s risk for contralateral cancer increases by 0.5-to-1.0 percent annually in the first ten years after diagnosis. Women diagnosed with atypical ductal hyperplasia (ADH) or lobular carcinoma in situ (LCIS) face a higher risk for development of breast cancer.

African-American women. The ACR and SBI are the first organizations to identify African-American women as being at high-risk for breast cancer. Although the diagnosis rates for black and white women are similar, newer data suggests that non-Hispanic black women experience a death rate that is 39
percent higher than non-Hispanic white women. Reasons vary and include:

- Access to breast cancer screening
- Access to breast cancer risk counseling
- Lack of healthcare insurance
- Tumor biology
- Higher incidence of triple-negative breast cancer
- Higher incidence of the BRAC1 and BRAC2 genetic mutations

A next-generation sequencing study of 289 African-American women revealed that 65 of them were positive for inherited mutations — a plausible reason for younger-onset, aggressive breast cancer in African-American women.

**Dense breasts.** More than 40 percent of women in the U.S. have dense breasts. They are classified as having heterogeneous dense breasts or extremely dense breasts. Women with extremely dense breasts face a 4X greater risk of breast cancer than women with fatty breasts. Most women’s breast tissue is somewhere in the middle between the extremes. It’s believed that these women who fall in the middle have a relative risk factor of 1.45.

**Current Mammography Guidelines**

**Mammography (preferably digital breast tomosynthesis, DBT)**

Women of Average Risk (<15%) with no family history of breast cancer or previous history of breast cancer.

- Begin mammography at age 40 and continue on an annual basis.

Woman at Increased Risk for Breast Cancer

- Women with BRCA1 or BRCA2 mutations and untested first-degree relatives of those who test positive for BRCA mutations — begin by age 30 (but not before age 25)
• Women with >20% lifetime risk based on family history (both maternal and paternal) – begin by 30 (but not before 25) or ten years earlier than the age diagnosed of the youngest-diagnosed relative, whichever is later
• Women whose mother or sisters diagnosed with pre-menopausal breast cancer – begin by 30 (but not before 25) or ten years earlier than the age diagnosed of the youngest-diagnosed relative, whichever is later
• Women with a history of chest or mantle radiation between the ages of 10 and 30 – annually beginning eight years after radiation treatment, but not before 25
• Women with proven diagnosis of atypical ductal hyperplasia (ADH) or lobular carcinoma in situ (LCIS) – annually, irrespective of age

Women should continue to have annual mammograms until their life expectancy is less than 5-to-7 years unless comorbidities would prevent action.

**Ultrasound (in addition to mammography)**

• Women who have dense breast tissue – to supplement/clarify results of digital breast tomosynthesis
• Women at high-risk who cannot have an MRI for whatever the reason

**MRI (in addition to mammography)**

• Women with a proven deleterious BRCA mutation – annually by age 30
• Untested first-degree relatives of a proven BRCA carrier – annually by age 30
• Women who have a >20% lifetime risk because of family history – annually starting by age 30
• Women with history of chest radiation – annually beginning eight years after radiation treatment
• Women recently diagnosed with breast cancer who have
normal contralateral breast by DBT and physical examination – MRI screening of the contralateral breast at diagnosis

- Optional – for women with a history of breast or ovarian cancer or history of ADH

**RECOMMENDATION:** All women should receive a risk assessment of their lifetime risk for breast cancer, with particular emphasis that every woman of African-American and Ashkenazi Jewish descent be assessed and identified by age 30 so that higher-risk women can begin appropriate screening by age 30.