High-risk women—What breast screening is appropriate?

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**Question**

Your 28-year-old patient has a first-degree relative with a known disease-causing BRCA1 or BRCA2 mutation, but she herself has not been tested. Which breast screening method(s) should you recommend?

**CHOOSE ONE:**

A. None until age 30; screening mammography/tomosynthesis alone at age 30 and beyond

B. Screening annual MRI alone until age 30; screening annual MRI and mammography/tomosynthesis at age 30 and beyond

C. Ultrasound

D. None; screening should not begin until age 40

**Answer**

**B.** For a 28-year-old woman, herself untested, with a known pathogenic BRCA1 or BRCA2 mutation in a first-degree relative (mother/father, sister/brother, daughter/son), screening annual magnetic resonance imaging (MRI) alone is recommended until age 30, followed by screening annual MRI and mammography/tomosynthesis at age 30 and beyond. If MRI is not an option, ultrasonography or contrast-enhanced mammography should be considered.

Most medical societies in the United States recommend mammography screening beginning at age 40 if a woman is at average risk. If a woman is determined to be at high risk, breast cancer screening may be recommended to begin by age 30. **Breast cancer risk assessment, with a risk model based largely on family history, should begin by age 30 for all women.**

The American College of Radiology (ACR), National Comprehensive Cancer Network (NCCN), and American Society of Breast Surgeons recommend annual MRI screening for the following high-risk subgroups of women:

- Women with known disease-causing BRCA1 or BRCA2 mutations, or other disease-causing mutations, or their untested first-degree relatives. **(Age to begin screening MRI and screening mammography/tomosynthesis varies by mutation)**

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The authors report that DenseBreast-info, Inc. is a 501(c)(3) nonprofit organization which receives unrestricted educational grants from GE Healthcare, Bayer, Volpara Health, Hologic, Siemens Healthineers, Beekey Medical, iCAD, and CMR Naviscan. Dr. Berg also reports being principal investigator for research from Koios Medical, Inc. for which her department receives a grant.

doi: 10.12788/obgm.0163

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adding annual digital mammography/tomosynthesis at age 30 and beyond (unless the woman has had bilateral mastectomy). Note: There is emerging evidence that the benefit of mammography is relatively small in pathogenic BRCA1 carriers prior to the age of 40; therefore, the ACR suggests BRCA1 mutation carriers may consider delaying mammography until age 40 only if they receive contrast-enhanced MRI annually starting at age 25.1 Annual mammography is of benefit beginning at age 30 in those with BRCA2 or other disease-causing mutations.

Age to begin annual MRI screening and mammography/tomosynthesis in women who are known to carry or are first-degree untested relatives of individuals with less common disease-causing mutations (such as those associated with Li-Fraumeni syndrome [TP53]; Bannayan-Riley-Ruvalcaba syndrome or Cowden syndrome [PTEN]; hereditary diffuse gastric cancer [CDH1]; Peutz-Jeghers syndrome [STK11]; neurofibromatosis type 1 [NF1]; PALB2; ATM; CHEK2; or BARD1) ranges from 20-40 years depending on the mutation and family history.3 (See https://densebreast-info.org/providers-faqs /what-is-the-screening-management-for -various-other-mutation-carriers/.)

• Women who received chest/mantle radiation therapy by age 30 (such as for Hodgkin disease) and at least 8 years prior. Women with prior chest radiation therapy (such as for Hodgkin disease) between ages 10 and 30 are at high risk for developing breast cancer;1,2,4,5 with risk similar in magnitude to BRCA1 or BRCA2 carriers, and are recommended for annual screening MRI starting at age 25 or 8 years after the chest radiation therapy, whichever is later.

• Women with a calculated lifetime risk of breast cancer of ≥20% are recommended to begin annual screening MRI by age 25-30.1,2,5 Any of the models that include detailed family history such as the Tyrrel-Cuzick (IBIS, which now includes breast density as a risk factor); BRCA PRO; BOADICEA; Claus; or Penn II; but not the Gail or Breast Cancer Surveillance Consortium (BCSC) models, can be used to estimate lifetime risk for the purposes of screening MRI guidelines. (See https://densebreast -info.org/for-providers/risk-model-tutorial/ for a summary table with live links.)

• Women with a personal history of breast cancer and dense breasts or diagnosis by age 50, regardless of breast density. A personal history of breast cancer is not included in risk models, but all women diagnosed with breast cancer at or before age 50 and treated with breast-conserving therapy have a ≥20% lifetime risk for a new breast cancer.1,2 Annual MRI may be considered in addition to annual mammography or tomosynthesis in women with a personal history of breast cancer diagnosed after age 50 and without dense breasts, and/or a history of lobular carcinoma in situ (LCIS) or prior atypia (eg, atypical ductal hyperplasia (ADH), atypical lobular hyperplasia (ALH), or atypical papilloma).1,2

Supplemental MRI screening should continue until age 75, after which management should be considered on an individual basis. If MRI screening is not an option, ultrasound or contrast-enhanced mammography (where available) should be considered as an alternative.6-8

References