

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),¹ National Comprehensive Cancer Network (NCCN),^{2,3} 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).**^{1,3} Women with known pathogenic *BRCA1* or *BRCA2* mutations, or their untested first-degree relatives, should begin annual screening with MRI only between ages 25-29,

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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 $\geq 20\%$** 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 Tyrer-Cuzick (IBIS, which now includes breast density as a risk factor); BRCAPRO; 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 $\geq 20\%$ 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 ●

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