Breast Cancer Risk Assessment Models and High-Risk Screening

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KEYWORDS

• Breast cancer screening • High-risk screening • Risk assessment models • Mammography • Breast MR imaging • Hereditary cancer syndromes

KEY POINTS

- The paradigm has recently shifted from a uniform method of breast cancer screening to an individualized approach that incorporates patient risk factors.
- Assessment of risk can be a complicated process and is greatly enhanced when a genetic counselor is involved to perform a comprehensive evaluation.
- Although the breast imaging center may be the logical location for this triage to occur, many patients with family history and elevated risk may benefit from earlier screening and require intervention before the age at which they present for routine breast screening.
- This article provides a practical approach to the risk assessment process and includes an overview of the risk models paired with recommendations for those at elevated risk for breast cancer.

INTRODUCTION

Significant progress has been made over the past few decades in the fight against breast cancer. Most notably, screening mammography has resulted in mortality reduction. Screening for breast cancer is performed in patients without signs or symptoms of disease with a goal of discovering the disease in an early stage when it is more treatable, less debilitating, and has improved survival rates. In fact, the stage at diagnosis influences overall survival significantly, regardless of advances in therapy.[1] There are, however, limitations to mammography, especially in women with increased risk for breast cancer because many of these women are younger and have dense breasts.

Screening recommendations vary based on patients' risk of developing the disease. Because of many factors, including hereditary risk, the incidence of breast cancer increases. Although mammography is the only imaging test that has proven to decrease breast cancer mortality, supplemental screening with additional imaging tools has shown increased cancer detection in patients particularly with an elevated risk of breast cancer. This finding began a movement to personalize or individualize breast cancer screening based on patient risk factors.

With new scientific evidence available regarding breast MR imaging in 2007, the American Cancer Society (ACS) published breast cancer screening recommendations that specifically included women at high risk for developing breast cancer. The ACS concluded that annual screening mammography and breast MR imaging may be indicated in patients with an elevated of risk of breast cancer.[2] Additionally, patients with elevated risk may also benefit from beginning screening earlier than those with average risk. This conclusion was largely based on research that found an increase in cancer detection with the addition of breast MR imaging or ultrasound to screening mammography.[3]

A thorough evaluation of individual risk factors will help establish personalized screening

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protocols. In addition, risk assessment will determine if patients are candidates for genetic testing or may benefit from chemoprevention.

BREAST CANCER RISK ASSESSMENT

Breast cancer risk assessment can be confusing because various tools and models exist. These tools and models can be broken down into 3 categories:

- Genetic counseling referral guidelines: A patient's personal/family history that is suspicious for a hereditary cancer syndrome will trigger a genetic counseling referral. At this point, the genetic counselor can gather a more detailed medical history and determine whether genetic testing is appropriate. Not everyone who is identified for referral to a genetic counselor will be a candidate for genetic testing.
- Genetic testing guidelines: These guidelines, often used by genetic counselors, are primarily designed to determine if genetic testing for a particular hereditary cancer syndrome is indicated.
- Breast cancer risk assessment models: These models are mathematical models that estimate a woman's risk to develop breast cancer over defined time periods. They are used to identify patients who can benefit from interventions, such as chemoprevention for risk reduction and breast MR imaging screening as an adjunct to mammography for increased surveillance. They are often not applicable to patients with a hereditary cancer syndrome, emphasizing why genetic counseling/testing should be pursued first when indicated.

Genetic Counseling Referral Guidelines

There are many models available to estimate a woman's risk to develop breast cancer over a specific time period. However, it is important to understand that many of these models are not applicable to women who carry a pathogenic variant (also known as a mutation) in a highly penetrant gene, such as *BRCA1* or *BRCA2*. In these situations, the presence of the pathogenic variant is often what defines the woman's risk to develop breast cancer and, depending on which gene is involved, may define her risk for other types of cancer as well. Thus, when embarking on breast cancer risk assessment, the health care provider must first determine whether patients meet the criteria for referral to a genetic counselor and/or discussion of genetic testing for hereditary cancer syndromes (**Fig. 1**).

The National Comprehensive Cancer Network's (NCCN) guidelines are the most commonly used parameters for identifying patients who are candidates for genetic counseling and testing. Their guidelines include criteria for referral to a genetic counselor for consideration of genetic testing.[4] Additional tools exist and are endorsed specifically by the United States Preventive Services Task Force (USPSTF) to determine the need for in-depth cancer genetic counseling for hereditary breast cancer (**Table 1**).[5]

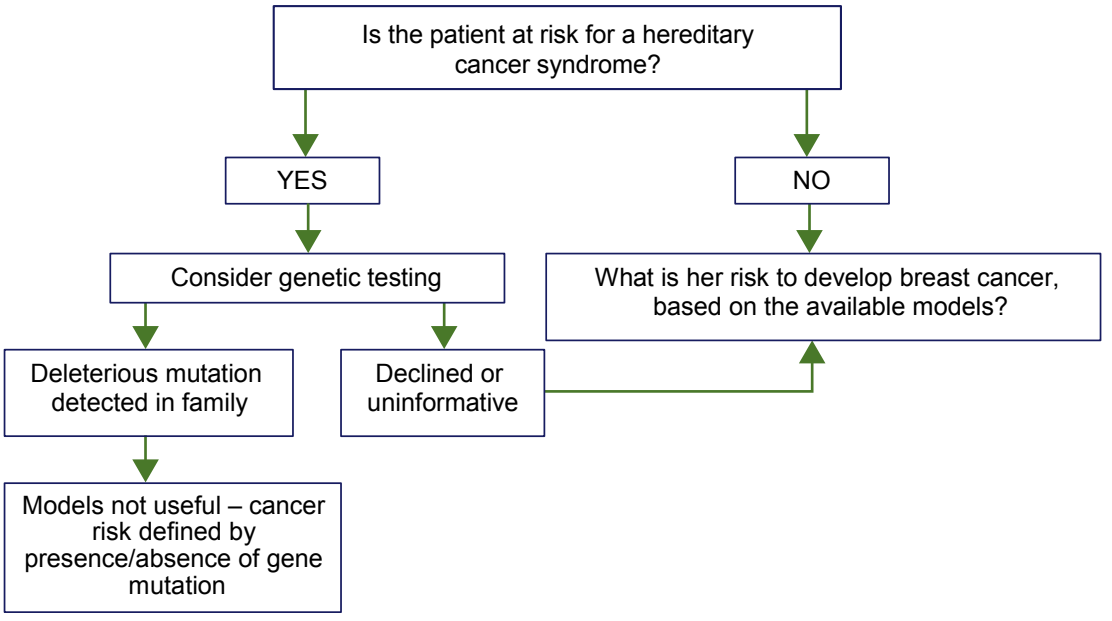

Fig. 1. Algorithm for breast cancer risk assessment.