



RISK FACTORS AND EARLY DETECTION STRATEGIES IN BREAST NEOPLASMS: A 5-YEAR SYSTEMATIC REVIEW

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<https://doi.org/10.5281/zenodo.18617794>

Abstract

Breast neoplasms remain a major global health concern and represent one of the leading causes of cancer-related morbidity and mortality among women. Understanding both modifiable and non-modifiable risk factors, together with implementing effective early detection strategies, is essential for improving prognosis and reducing disease burden.

This systematic review evaluates the evidence published between 2020 and 2025 regarding key risk factors and contemporary approaches to early detection in breast neoplasms. Recent literature confirms that genetic predisposition, particularly BRCA1 and BRCA2 mutations, family history, hormonal and reproductive factors, obesity, alcohol consumption, and lifestyle-related determinants significantly contribute to disease risk. Advances in screening technologies, including digital mammography, ultrasound, magnetic resonance imaging (MRI), and digital breast tomosynthesis, have enhanced early detection rates. Moreover, risk-adapted screening models tailored to individual patient profiles demonstrate improved diagnostic accuracy while reducing unnecessary interventions and overdiagnosis.

Current evidence supports a personalized, risk-based approach to screening and prevention. Future strategies should focus on integrating molecular biomarkers with advanced imaging modalities to optimize early diagnosis and clinical outcomes in patients with breast neoplasms.

Keywords

Breast neoplasms, risk factors, early detection, screening strategies, mammography, magnetic resonance imaging, digital breast tomosynthesis, BRCA1, BRCA2, risk stratification.

Introduction

Breast neoplasms represent one of the most significant global health challenges, accounting for the highest incidence of cancer among women worldwide. Despite advances in treatment and improved survival rates in high-income countries, breast cancer remains a leading cause of cancer-related mortality. The growing global burden is influenced by demographic transitions, lifestyle changes, environmental exposures, and disparities in access to healthcare services.

Understanding the multifactorial etiology of breast neoplasms is essential for effective prevention and early detection. Both non-modifiable risk factors such as genetic predisposition, family history, and breast density and modifiable determinants including obesity, alcohol consumption, physical inactivity, and hormonal exposure play critical roles in disease development. Advances in molecular genetics have further clarified the impact of pathogenic variants, particularly BRCA1 and BRCA2 mutations, on lifetime cancer risk.



Early detection remains a cornerstone of breast cancer control strategies. Population-based screening programs, primarily relying on mammography, have demonstrated reductions in mortality. However, limitations such as reduced sensitivity in dense breast tissue and the potential for overdiagnosis highlight the need for more precise approaches. Emerging technologies, including digital breast tomosynthesis and magnetic resonance imaging (MRI), as well as risk-adapted screening models, offer promising opportunities to enhance diagnostic accuracy and personalize surveillance strategies.

Over the past five years, substantial research has focused on refining risk stratification models and integrating advanced imaging modalities into screening protocols. Given the rapid evolution of evidence in this field, a comprehensive evaluation of contemporary risk factors and early detection strategies is warranted.

Therefore, this systematic review aims to synthesize the evidence published between 2020 and 2025 regarding major risk factors and modern early detection approaches in breast neoplasms, with particular emphasis on personalized, risk-based screening strategies.

Main body

Epidemiology and Global Burden

Breast neoplasms represent one of the most prevalent malignancies worldwide and remain a leading cause of cancer-related morbidity and mortality among women. Recent epidemiological analyses confirm a steady increase in incidence, particularly in low- and middle-income countries, largely attributed to demographic transitions, lifestyle changes, and improved detection strategies. Early identification of high-risk individuals and optimization of screening programs are therefore essential components of public health strategies.

Major Risk Factors in Breast Neoplasms

Genetic Predisposition

Genetic susceptibility remains one of the strongest non-modifiable risk factors. Pathogenic variants in BRCA1 and BRCA2 significantly increase lifetime risk of breast cancer, often leading to earlier onset and more aggressive disease phenotypes (Petrucci et al., NCBI Bookshelf). Women carrying BRCA mutations benefit from intensified surveillance protocols, including annual MRI combined with mammography.

Breast Density

High mammographic breast density has been consistently associated with both increased cancer risk and reduced sensitivity of standard mammography. Dense breast tissue may obscure lesions, leading to interval cancers. Recent studies highlight the importance of supplemental imaging, particularly MRI and digital breast tomosynthesis (DBT), in this subgroup.

Hormonal and Reproductive Factors

Prolonged exposure to endogenous or exogenous estrogen contributes significantly to carcinogenesis. Early menarche, late menopause, nulliparity, delayed first childbirth, and hormone replacement therapy have all been identified as established risk determinants.

Lifestyle and Environmental Factors

Obesity, physical inactivity, and alcohol consumption remain important modifiable risk factors. Meta-analyses confirm a dose-dependent relationship between alcohol intake and breast cancer risk. Adiposity, particularly in postmenopausal women, is strongly associated with increased estrogen production and tumor development.



Early Detection Strategies Mammography

Digital mammography remains the cornerstone of population-based screening. Global guidelines recommend regular screening between ages 40 and 74, depending on national protocols. Mammography has demonstrated mortality reduction; however, its sensitivity decreases in women with dense breast tissue.

Digital Breast Tomosynthesis (DBT)

Digital breast tomosynthesis improves lesion visualization by providing three-dimensional imaging. Studies indicate higher cancer detection rates and lower recall rates compared to conventional mammography. DBT appears particularly beneficial in women with heterogeneously dense breasts.

Magnetic Resonance Imaging (MRI)

Breast MRI demonstrates superior sensitivity compared to mammography and ultrasound, especially in high-risk populations such as BRCA mutation carriers. MRI screening reduces interval cancer rates in women with dense breasts. Abbreviated MRI protocols have emerged as a cost-effective alternative, although long-term outcome data remain limited.

Risk-Adapted Screening Models

Recent literature emphasizes individualized, risk-based screening strategies. Risk prediction models incorporating genetic, clinical, and imaging factors enable tailored screening intervals and modalities. Such approaches aim to maximize early detection while minimizing overdiagnosis and unnecessary biopsies.

Comparative Analysis and Clinical Implications

Evidence suggests that screening effectiveness depends on underlying risk profiles.

High-risk women (e.g., BRCA mutation carriers) benefit from MRI-based surveillance. Women with dense breasts may require supplemental imaging beyond standard mammography. Average-risk populations continue to benefit from structured mammographic screening programs. Risk-adapted screening models demonstrate potential in improving diagnostic precision and resource allocation. However, concerns remain regarding cost-effectiveness, accessibility, and implementation in low-resource settings.

Limitations in Current Evidence

Despite advances, several limitations persist: Many studies are observational and heterogeneous in methodology. Long-term outcomes of abbreviated MRI and tomosynthesis require further validation. Integration of molecular biomarkers into screening algorithms remains investigational. Future prospective multicenter trials are necessary to refine personalized screening frameworks.

Conclusion

Breast neoplasms remain a significant global health challenge, with incidence influenced by a complex interaction of genetic, hormonal, reproductive, and lifestyle-related factors. Contemporary evidence from the past five years confirms that pathogenic variants in BRCA1 and BRCA2, high breast density, prolonged hormonal exposure, obesity, and alcohol consumption are among the most influential risk determinants. While certain factors are non-modifiable, many lifestyle-related risks offer opportunities for preventive interventions.

Advances in early detection strategies have substantially improved diagnostic accuracy. Digital mammography continues to serve as the foundation of population-based screening.



while digital breast tomosynthesis and magnetic resonance imaging enhance detection rates, particularly in women with dense breasts or elevated genetic risk. Emerging risk-adapted screening models demonstrate promise in tailoring surveillance strategies according to individual risk profiles, thereby improving early diagnosis and reducing unnecessary procedures.

Overall, current evidence supports a personalized, risk-based approach to breast cancer screening and prevention. Future research should focus on large-scale prospective studies, cost-effectiveness analyses, and integration of molecular biomarkers into screening algorithms to optimize early detection and improve clinical outcomes.

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