

Early diagnosis and prevention of breast diseases: From mammography screening to new technologies. A Literature Review

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DOI: <https://doi.org/10.5281/zenodo.19759866>

Published Date: 25-April-2026

Abstract: Early diagnosis and prevention of breast diseases is a critical component of public health, particularly in addressing breast cancer, which remains one of the most prevalent cancers affecting women worldwide. The paradigm of breast cancer screening has largely revolved around mammography, a technique notable for its ability to detect cancer at an early stage, thereby improving survival rates when coupled with effective treatment. Despite its proven benefits, mammography is not without limitations, including the potential for false positives and overdiagnosis, particularly in certain age groups. As the field evolves, innovative technologies and approaches are emerging, aiming to enhance early detection and individual risk assessment, such as the consideration of mammographic density and its significant correlation to breast cancer risk. Moreover, the necessity for tailored screening programs that accommodate diverse population needs has been emphasized in global initiatives, advocating for resource-stratified guidelines that improve access and efficacy across various healthcare settings. This review seeks to explore these advancements and their implications for breast disease prevention.

Keywords: breast cancer, early diagnosis, prevention, mammography, risk assessment.

1. INTRODUCTION

Breast diseases, particularly breast cancer, represent a significant public health challenge globally, affecting millions of women each year. The significance of early detection cannot be overstated, as breast cancer diagnosed in its initial stages is highly treatable, resulting in improved survival rates. Mammography stands as the cornerstone of breast cancer screening, facilitating early diagnosis and offering a pathway to effective interventions. However, screening practices must consider various factors, including the age of women and associated risks. For instance, while mammography is beneficial, it also presents challenges such as false positives and overdiagnosis, especially in women aged 40 to 49 years (1). Furthermore, recent findings underscore the importance of mammographic density in evaluating breast cancer risk, suggesting that this parameter could guide personalized screening strategies (2). The introduction of artificial intelligence into mammography interprets these data effectively, enhancing accuracy in lesion detection and diagnosis while addressing workflow issues inherent in clinical settings (3). However, barriers to access exist that can hinder effective breast health care, particularly in resource-limited settings, highlighting the need for comprehensive early detection programs (4).

Early diagnosis and prevention of breast diseases are paramount to improving survival rates and reducing treatment-related morbidity. Studies have consistently shown that when breast cancer is detected at an early stage, patients have significantly

better prognoses, often resulting in higher survival rates. For instance, Ginsburg (4) emphasizes that early detection not only enhances survival probabilities but also emphasizes the necessity of addressing systemic barriers that may hinder access to effective screening. Mammography has long been the cornerstone of breast cancer screening, though its effectiveness can vary across age groups, underscoring the necessity for tailored approaches in different demographics (1). Moreover, advancements in technology, particularly the integration of artificial intelligence into screening processes, are enhancing diagnostic accuracy and efficiency, with emerging algorithms showing potential in lesion detection and density assessment (3). Collectively, these advancements speak to a broader goal of fostering preventative strategies that are both accessible and effective, ensuring that more women benefit from timely interventions and a comprehensive understanding of their risks (2).

In establishing the objective and scope of the literature review on early diagnosis and prevention of breast diseases, it becomes crucial to synthesize existing research while illuminating the gaps that need addressing. The review reveals that mammography remains a pivotal tool in breast cancer screening but has limitations in its effectiveness across different age groups, particularly in women aged 40 to 49 years, where benefits may be counterbalanced by risks such as overdiagnosis and false positives (1). Furthermore, it highlights significant correlations between mammographic density and breast cancer risk, emphasizing the need for innovative screening methods that prioritize personalized approaches (2). Addressing broader socio-economic barriers to healthcare access is essential for effective implementation of early detection programs (4). Ultimately, the review aims to explore emerging technologies and frameworks that facilitate personalized interventions, thereby enhancing early detection strategies and improving health outcomes (5).

The landscape of breast disease diagnosis has evolved significantly with the introduction of various methodologies and technologies that enhance early detection and prevention strategies. Traditional mammography remains a cornerstone for screening, yet its effectiveness is often hindered by variability in human interpretation, leading to inconsistent diagnoses. Recent advancements incorporate Artificial Intelligence (AI) applications to augment the interpretative domains of mammography, such as lesion detection and triage, which can significantly improve diagnostic accuracy while reducing false positives (4). Additionally, research indicates a strong correlation between mammographic density and breast cancer risk, underscoring the clinical importance of integrating this factor into screening protocols (2). The Breast Health Global Initiative emphasizes that addressing barriers to early detection—ranging from socio-economic issues to access to healthcare services—is crucial for successful implementation of screening programs across different resource settings (3). These developments highlight a multifaceted approach to breast disease management, enhancing prospects for early diagnosis and improved patient outcomes (6).

Mammography Screening in Breast Disease Diagnosis

Mammography screening serves as a cornerstone in the early diagnosis and prevention of breast diseases, particularly breast cancer, which remains a prevalent health concern for women globally. The efficacy of mammography in reducing mortality rates is well-documented, emphasizing its critical role in the timely identification of tumors that may not yet exhibit clinical symptoms. As indicated in current literature, mammographic density has emerged as a significant factor in assessing individual breast cancer risk, underscoring the need for nuanced screening protocols that account for this variable (2). Additionally, variations in the benefits and risks associated with mammography across different age demographics highlight the complexity of its effectiveness, particularly for women aged 40 to 49, who may experience a higher incidence of false positives and overdiagnosis (1). The identification of benign breast disease via mammography has been linked to an increased long-term risk of developing breast cancer, further necessitating personalized screening approaches to enhance early detection strategies (7). Despite ongoing challenges related to access and healthcare equity, developing innovative and resource-stratified screening programs is essential for maximizing early diagnosis opportunities (4).

The history and development of mammography have significantly transformed the landscape of breast cancer detection and diagnosis. Initially introduced in the mid-20th century, mammography evolved from basic radiographic techniques to sophisticated imaging modalities that enhance sensitivity and specificity. This evolution is critical, as mammography is a cornerstone in breast cancer screening, reducing mortality through early detection (1). Advances in technology, such as digital mammography and tomosynthesis, have further improved diagnostic accuracy, allowing healthcare providers to identify denser breast tissues that may obscure lesions (2). The importance of timely screening is underscored by studies emphasizing that early diagnosis can drastically improve survival rates, as any diagnostic delay can adversely affect patient

outcomes (8). However, substantial barriers to access remain in various settings, including socioeconomic factors that hinder women's timely printing and follow-ups (4). Moving forward, the continued refinement of mammographic techniques, alongside comprehensive programs addressing these barriers, will be essential to bolster early detection efforts and enhance breast cancer outcomes globally.

Traditional mammography has long been regarded as a cornerstone in the early detection and prevention of breast cancer, demonstrating significant efficacy in reducing mortality rates among women, particularly in ages 40 to 49 years (1). However, its limitations cannot be overlooked. While mammography enhances early detection, it is also associated with drawbacks such as false positives, overdiagnosis, and exposure to radiation, which may lead to unnecessary anxiety and invasive procedures (2). Moreover, mammographic density becomes a critical factor; higher densities are linked to an increased risk of developing breast cancer, thus necessitating the exploration of personalized screening strategies to improve outcomes (2). As highlighted in recent discussions surrounding the European Collaborative on Personalized Early Detection of Breast Cancer, there is a pressing need for innovative approaches to screening that address the diverse risk profiles among women. Ultimately, while traditional mammography remains a vital tool, integrating advanced technologies and personalized risk models is essential for optimizing breast cancer detection and management (4).

Effective screening guidelines and population-based programs are pivotal in the early diagnosis and prevention of breast diseases, particularly through mammography and emerging technologies. Research demonstrates that while mammography significantly reduces mortality rates, its effectiveness varies across age groups, necessitating adaptable screening strategies tailored to specific populations (1). In Malaysia, for instance, analyses of national breast cancer screening initiatives highlighted fluctuations in participation rates, influenced by health promotion campaigns, which underscore the vital role of community engagement in enhancing screening uptake (9). Furthermore, the Breast Health Global Initiative has emphasized the importance of resource-stratified guidelines that account for socio-economic barriers, thereby providing a framework for implementing successful early detection programs in diverse settings (4). Developing personalized risk assessment tools and addressing ethical considerations are similarly critical, as noted in the ENVISION consensus conference, to create effective evidence-based interventions in breast cancer screening (5). These guidelines unite efforts toward improving early diagnostic practices globally.

Breast cancer continues to pose significant public health challenges worldwide, significantly impacting mortality rates and the effectiveness of detection strategies. Early detection through screening methods such as mammography has been shown to reduce breast cancer mortality, yet the benefits vary among different age groups. In particular, women aged 40 to 49 may experience heightened risks associated with mammography, including false positives and overdiagnosis, which complicate the assessment of its overall effectiveness (5). Furthermore, mammographic density has emerged as a critical factor in screening accuracy and individual risk assessment, indicating that variations in breast tissue composition can influence cancer risk (1). Innovative approaches, such as those proposed by the ENVISION network, aim to refine risk prediction methods and develop personalized interventions that account for an individual's ancestry and breast cancer subtype (2). These advancements will be vital in not only enhancing detection rates but also in addressing the disparities in breast cancer outcomes among diverse populations (10).

Advances in Imaging Technologies for Breast Disease

The evolution of imaging technologies has markedly transformed the landscape of breast disease diagnosis, enhanced early detection and potentially improved patient outcomes. While mammography remains the cornerstone for breast cancer screening, its limitations have prompted significant advancements in alternative modalities. Digital breast tomosynthesis (DBT) has emerged as a promising solution, offering three-dimensional imaging that improves diagnostic accuracy and reduces rates of false positives, as highlighted by recent literature reviews (8). Furthermore, innovations such as contrast-enhanced mammography and magnetic resonance imaging have expanded the diagnostic toolkit, providing clinicians with detailed images that facilitate the assessment of complex cases (11). The integration of artificial intelligence and machine learning into these imaging techniques represents a crucial frontier, helping to interpret images with greater precision and consistency (12). Meanwhile, ongoing research into the relationship between mammographic density and breast cancer risk underscores the need to tailor screening approaches based on individual patient profiles (2). Collectively, these technological advances are not only refining diagnostic processes but also fostering a paradigm shift towards more personalized and effective breast health management.

The evolution of breast cancer screening technologies has underscored the significance of digital mammography (DM) and digital breast tomosynthesis (DBT) in improving early diagnosis and prevention of breast diseases. While mammography has long served as the cornerstone of screening efforts, concerns about its limitations, particularly in women with dense breast tissue, have prompted the exploration of DBT as a complementary modality. Studies indicate that DBT enhances the sensitivity of breast cancer detection, allowing for clearer visualization of overlapping structures, which is particularly beneficial in complex breast compositions (13). It is also associated with a reduction in false-positive rates, ultimately mitigating unnecessary anxiety and procedures for patients (11). Furthermore, the implementation of artificial intelligence in conjunction with these imaging techniques holds promise for refining risk assessment and optimizing the application of DBT, potentially guiding its use for women most likely to benefit from it (14). Initiatives like the Malmö Breast Imaging database are crucial for supporting ongoing research and validation of these technologies, ensuring that advancements in diagnostic imaging lead to improved patient outcomes (14).

The integration of advanced imaging techniques such as ultrasound and magnetic resonance imaging (MRI) has significantly enhanced the early diagnosis and prevention of breast diseases. While conventional mammography remains a standard screening method, its limitations—particularly in women with dense breast tissue—have prompted the exploration of alternative modalities. Ultrasound, specifically automated breast ultrasound (ABUS), offers improved diagnostic accuracy by effectively visualizing dense tissues, thereby facilitating the early detection of malignancies (15). MRI, with its high sensitivity, plays a crucial role in risk assessment and pre-operative evaluation, particularly for high-risk populations (11). Additionally, with artificial intelligence (AI) technologies being integrated into these imaging techniques, the potential for enhancing diagnostic precision is substantial, as AI applications in medical imaging improve lesion identification and classification (16). Collectively, these technologies not only mitigate some of the drawbacks associated with traditional mammography, but also pave the way for a more accurate and comprehensive approach to breast cancer screening and diagnosis in clinical practice (1).

As breast cancer remains one of the leading causes of mortality among women globally, advancements in imaging techniques are critical to enhancing early diagnosis and prevention efforts. Among these innovations, contrast-enhanced mammography (CEM) stands out due to its promising utility in oncological diagnostics. CEM utilizes an iodine-based contrast agent in conjunction with standard mammographic techniques, allowing for improved identification of tumor angiogenesis, akin to dynamic contrast-enhanced magnetic resonance imaging (DCE-MRI) (8). This method not only exploits the sensitivity of full-field digital mammography (FFDM) but also provides additional insights regarding tumor morphology and function (11). The evidence suggests that CEM may approach the diagnostic efficacy of DCE-MRI while offering a more accessible and less costly alternative (17). Given the potential for CEM to reduce diagnostic delays, particularly in under-resourced settings post-pandemic, its implementation could significantly impact breast cancer outcomes (12). Thus, embracing such technologies is essential for improving early detection strategies.

The integration of artificial intelligence (AI) into imaging interpretation presents transformative potential for the early diagnosis and prevention of breast diseases, particularly through mammography screening. Various studies highlight that AI systems can enhance the detection of breast cancer by applying Clinical Decision Support System (CDSS) technologies, which leverage machine learning to assist radiologists in identifying cancerous lesions more accurately and consistently (6). AI techniques excel in medical imaging, demonstrating the capacity for lesion identification, segmentation, and classification, thereby effectively improving screening outcomes (16). Notably, efforts are underway to validate AI systems against entire screening populations, providing comparative insights into their efficacy alongside human radiologists (18). Furthermore, current commercial applications of AI not only focus on lesion detection but also encompass risk assessment and image quality enhancement, highlighting their multifaceted utility in clinical practice (3). Hence, the potential benefits of AI systems in imaging interpretation represent a significant advancement in the early diagnosis of breast cancer, ultimately improving patient outcomes.

Emerging Techniques and Preventive Strategies

In the evolving landscape of breast disease management, emerging techniques and preventive strategies are pivotal in advancing early diagnosis and improving patient outcomes. Traditional mammography, while beneficial for detecting breast cancer, reveals its limitations particularly regarding mammographic density, which significantly correlates with breast cancer risk and necessitates innovative approaches in screening protocols (2). Research emphasizes that personalized risk

assessment tools, tailored to diverse populations, enhance early detection efforts by addressing individual risk factors and potentially increasing utilization of screening services, especially among women aged 40 to 49 (1). Furthermore, the integration of advanced imaging technologies, such as MRI and ultrasound, along with new technologies for non-surgical preventive measures, can provide broader options for managing high-risk patients (5). The development of resource-stratified guidelines by initiatives like the Breast Health Global Initiative illustrates the importance of addressing systemic barriers to access, ultimately improving early detection programs worldwide (4).

The integration of biomarkers and molecular diagnostic tools has emerged as a transformative advancement in the early diagnosis and prevention of breast diseases. These tools not only enhance traditional screening methods but also facilitate a more personalized approach to cancer management. For instance, the application of circulating tumor DNA (ctDNA) analysis allows for non-invasive monitoring of tumor dynamics, complementing conventional imaging techniques like mammography. Recent literature emphasizes the importance of these biomarkers in detecting breast cancer at earlier stages, thus improving treatment outcomes and survival rates. Additionally, the development of advanced technologies, such as artificial intelligence algorithms, has the potential to analyze complex biomarker data more effectively, further aiding in the identification of at-risk populations (19). As the heterogeneity of breast cancer becomes increasingly understood, optimizing these molecular tools can significantly contribute to targeted therapeutic approaches, thereby addressing the diverse needs of patients (11). Ultimately, the ongoing evolution of biomarkers and molecular diagnostics offers unprecedented opportunities for enhancing breast cancer prevention strategies (20).

The integration of genetic screening and risk assessment models has revolutionized the landscape of breast cancer prevention and early diagnosis. By identifying genetic predispositions, such as BRCA mutations, these models provide crucial insights that enable personalized screening strategies tailored to individual risk profiles. Recent literature highlights that these genetic factors, when combined with traditional methods like mammography, significantly enhance prognostic accuracy and can guide preventive measures, ultimately reducing mortality rates (21). Evidence indicates that risk assessment tools that incorporate genetic data are essential for developing targeted interventions, particularly for women with a familial history of breast cancer. Furthermore, advancements in research are pushing for the establishment of breast cancer subtype-specific risk assessment tools that are applicable across diverse populations, addressing the ethical implications and fostering inclusivity in research (5). This multi-faceted approach not only underscores the importance of preventive measures but also emphasizes the necessity of ongoing research to refine these models, as they are pivotal in designing effective early detection strategies that can mitigate the impact of breast cancer (1,2).

Understanding the role of lifestyle interventions and chemoprevention is vital for enhancing preventative strategies against breast disease, particularly as early diagnosis becomes increasingly crucial. Emerging evidence underscores the impact of lifestyle choices—such as diet, physical activity, and weight management—on breast cancer risk. For example, certain dietary patterns rich in fruits and vegetables have demonstrated potential in reducing the incidence of breast cancer. Moreover, chemo-preventive agents, including selective estrogen receptor modulators and aromatase inhibitors, are being studied for their ability to mitigate risk among high-risk populations, including those with BRCA mutations. Furthermore, the integration of educational programs led by healthcare professionals, such as pharmacists, can significantly promote awareness about these interventions, ultimately encouraging women to adopt healthier lifestyles and participate in regular screening programs (22). As highlighted by the European Collaborative on Personalized Early Detection and Prevention of Breast Cancer, advancing personalized interventions that combine lifestyle modifications with chemoprevention will enhance the effectiveness of existing breast health strategies (5).

The integration of personalized medicine into the prevention of breast diseases represents a transformative shift toward more effective healthcare strategies. Personalized medicine leverages individual genetic, environmental, and lifestyle factors to tailor screening and prevention approaches, thereby enhancing early diagnosis and optimizing treatment outcomes. For instance, mammography remains a critical tool for detecting breast cancer; however, its efficacy can vary significantly among women, especially those aged 40 to 49 years (1). This highlights the necessity for risk stratification where mammographic density serves as a significant prognostic factor (2). Moreover, personalized interventions can also harness genetic testing to guide clinical decisions, ensuring that women receive tailored treatments aligned with their specific breast cancer subtypes (5). Ultimately, the alignment of personalized approaches with existing screening protocols aims to improve early detection while mitigating potential harms associated with conventional methods, refining the preventative landscape of breast health.

2. CONCLUSION

In conclusion, the early diagnosis and prevention of breast diseases represent significant advancements that have the potential to save countless lives. Mammography screening has long been the cornerstone of breast cancer detection, reducing mortality rates by facilitating timely intervention; however, its efficacy varies among different age groups, particularly in younger women where the benefits and risks must be carefully weighed (1). Furthermore, the association between mammographic density and breast cancer risk emphasizes the need for refined screening techniques that consider individual patient profiles, thereby enhancing diagnostic accuracy and preventive strategies (2). Research findings reveal that benign breast disease can substantially increase breast cancer risk over time, suggesting that those with such a diagnosis may require more intensive surveillance and personalized screening approaches (23). However, significant barriers to timely detection persist, influenced by social and economic factors that limit access to essential healthcare services (4). The ongoing development of innovative technologies in breast disease detection and the improvement of existing screening protocols will be critical in making prevention strategies more effective, ultimately leading to better health outcomes for women globally. Continuous efforts toward refining these practices are imperative to address the multifaceted challenges in breast health care.

A synthesis of the literature reveals significant advances and challenges in the early diagnosis and prevention of breast diseases, particularly concerning mammography screening. Effective mammography is noted for its vital role in reducing breast cancer mortality, yet its effectiveness varies across different age groups, particularly among women aged 40 to 49 years who may experience higher risks of false positives and overdiagnosis (1). Furthermore, studies have established a strong correlation between mammographic breast density and the risk of developing breast cancer, emphasizing the need for customized screening protocols that consider individual risk factors (2). Additionally, benign breast disease is highlighted as a long-term predictor of breast cancer risk, necessitating enhanced surveillance among affected women (23). Lastly, barriers to early detection are notably complex, encompassing social and economic factors that hinder access to necessary healthcare services, thereby underscoring the need for tailored intervention strategies (4). These findings collectively advocate for improved screening methodologies and public health strategies to enhance breast disease prevention.

Despite advancements in technologies and screening methodologies, early diagnosis and prevention of breast diseases encounter significant challenges and gaps that hinder effective patient outcomes. One primary issue lies in the variability of mammography's effectiveness across different age groups, particularly the debate surrounding its benefits versus risks in women aged 40 to 49, where overdiagnosis and false positives present serious concerns (1). Furthermore, as artificial intelligence (AI) continues to emerge as a transformative tool in breast cancer detection, the current evidence supporting its clinical applications remains largely insufficient, necessitating rigorous evaluation to ensure reliable integration into practice (3). Additionally, socio-economic and geographic barriers persistently obstruct access to timely screenings and preventive measures, highlighting the urgent need for resource-stratified guidelines that consider local contexts for program implementation (4). The development of personalized prevention strategies, which include subtype-specific risk assessment and novel interventions, is crucial for adapting to diverse populations and ensuring both effectiveness and ethical adherence in breast health care (5).

As the landscape of breast disease diagnosis continues to evolve, future directions in research must address the multifaceted challenges associated with early detection and prevention methods. A critical area of focus is the development and implementation of personalized screening strategies that account for genetic, socioeconomic, and demographic variations among women, particularly in resource-limited settings. The Breast Health Global Initiative emphasizes the necessity of resource-stratified guidelines for effective early detection programs, underscoring the importance of addressing complex barriers that women face, such as access to affordable care and education (4). Additionally, there is a significant opportunity for enhancing mammography screening through the integration of artificial intelligence technologies. These innovations can improve lesion detection and diagnostic accuracy, yet their clinical evaluation remains limited, necessitating further research into their effectiveness and ethical considerations in practice (3). Moreover, the European Collaborative on Personalized Early Detection and Prevention of Breast Cancer has identified urgent research priorities, including developing subtype-specific risk assessment tools and exploring novel preventive measures to decrease breast cancer incidence (5). Such interdisciplinary approaches present a promising trajectory for enhancing early diagnosis and prevention in breast health.

The implications for clinical practice and public health policy in the early diagnosis and prevention of breast diseases are profound, particularly as research evolves towards personalized interventions. The current reliance on mammography, while beneficial in detecting breast cancer, highlights a need for a more nuanced understanding of risks associated with screening guidelines, especially for women aged 40 to 49, as this age group exhibits varying benefits and risks depending on individual health profiles (1). Furthermore, insights into mammographic density have emerged as crucial for risk assessment, emphasizing the necessity for tailored screening strategies that account for this factor in clinical applications (2). The ENVISION initiative underscores the importance of developing subtype-specific risk tools and novel preventive measures that could significantly enhance the efficacy of screening programs (5). Consequently, addressing barriers to access, as noted by the Breast Health Global Initiative, is essential for ensuring equitable care and improving early detection outcomes across diverse populations (4).

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