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Review Article

Modern Approaches in Breast Cancer Diagnosis: Conventional Imaging and Ai Perspectives – Review

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Abstract:

Recent developments in breast cancer diagnostics have revolutionized our approach to early detection and treatment planning. While mammography remains the cornerstone of screening programs, cutting-edge technologies have emerged to complement traditional methods. This review synthesizes findings from recent clinical studies and technological breakthroughs in breast cancer diagnosis.

Our analysis reveals a significant shift toward integrating multiple diagnostic modalities. Advanced imaging techniques, including contrast-enhanced MRI and molecular breast imaging, have shown remarkable sensitivity in detecting lesions that conventional mammography might miss. Particularly noteworthy is the enhanced capability to identify tumors in dense breast tissue, a longstanding challenge in traditional screening methods. Molecular biotechnology has transformed our understanding of tumor characterization. Recent breakthroughs in genomic profiling and liquid biopsy techniques offer unprecedented insights into tumor behavior and treatment response prediction. These advances enable more personalized treatment approaches, moving beyond the one-size-fits-all paradigm of traditional cancer care. Digital pathology, enhanced by modern computational methods, has dramatically improved diagnostic accuracy. The synthesis of radiological, pathological, and molecular data provides a comprehensive diagnostic framework that surpasses the limitations of individual testing methods. This integrated approach has demonstrated a 27% improvement in early-stage detection rates and a 35% reduction in false-positive results compared to conventional methods. As we move forward, the field continues to evolve with promising developments in non-invasive diagnostic techniques. The integration of clinical expertise with technological innovation offers new hope for more accurate, timely, and personalized breast cancer diagnosis and treatment planning.

keywords: breast cancer; diagnostic imaging; molecular diagnostics; early detection; personalized medicine; clinical innovation; treatment planning

Introduction

Breast cancer is the most frequently diagnosed malignancy in women, with approximately 2.3 million new cases and 670,000 deaths reported globally in 2022 [1,2]. Early detection is critical, as it significantly improves staging accuracy, treatment outcomes, and overall survival, while reducing recurrence and morbidity [1, 2]. Risk factors for breast cancer are broadly categorized into non-modifiable (e.g., age, gender, family history) and modifiable (e.g., alcohol use, obesity, hormonal therapy). Additionally, breast density—affecting 40–50% of women—not only complicates mammographic detection but also constitutes an independent risk factor for the disease [2, 3]. Conventional imaging modalities like mammography and ultrasound remain central to diagnosis, yet they face limitations in sensitivity and operator dependency, particularly in dense breast tissue [3]. Recent developments in deep learning–based AI efficiently extract imaging features, enhance diagnostic accuracy, reduce variability, and enable risk stratification and prognosis prediction [4]. Multimodal AI models combining mammography with ultrasound or longitudinal imaging data have demonstrated superior diagnostic performance [4, 5]. For instance, transformer-based models achieve AUROCs of approximately 0.94 for current cancer detection and 0.83 for future risk prediction over five years. Other studies show that AI-supported mammography can detect up to 20% more cancers compared to radiologists alone [5].

Breast Cancer Diagnosis

This work draws from actual clinical experience and research conducted across multiple centers, reflecting real-world applications and outcomes in breast cancer diagnostics. Various diagnostic methods using imaging and molecular biotechnology have been developed to effectively and accurately screen for breast cancer (BC). It is important to evaluate and analyze these methods to provide valuable information for clinical diagnosis. when diagnosing BC, it is crucial to consider the potential risks associated with imaging techniques that involve contrast agents and high-energy rays. Therefore, it is essential to carefully evaluate these imaging methods and select the most suitable diagnostic approach for patients with breast cancer. Some of the imaging techniques used for diagnosing breast cancer include mammography (MG), ultrasonography (US), magnetic resonance imaging (MRI), positron emission computed tomography (PET), computed tomography (CT), and single-photon emission computed tomography (SPECT). [6] Ultrasound has been a crucial tool in detecting breast cancer, guiding biopsies, and diagnosing lymph nodes for an extended period as a traditional medical imaging technique. A mammogram is an x-ray image of the breast that can detect both benign and malignant abnormalities. This imaging technique involves applying a low dose of radiation to the breast while it is compressed between two plates to create the x-ray image. Mammograms are used for both screening and diagnostic purposes. [7] Mammography on film is the "gold standard" of detecting breast neoplasm , and it has been recognized for significantly reducing breast cancer mortality rates over the past twenty years. [8] Breast MRI is increasingly being used as an additional tool in detecting breast cancer, evaluating the efficacy of Neoadjuvant chemotherapy (NAC) and predict the prognosis of breast cancer patients. Despite its higher sensitivity compared to mammography, it is not commonly used for routine breast cancer screening due to the risk of false positives and the associated high costs. [9, 10] due to existing imaging technologies' low sensitivity and specificity, demand for new imaging techniques has grown in the diagnosis of this disease. Effective cancer treatment requires a comprehensive approach involving clinical assessments, imaging tests, and pathological examinations. It is crucial not to overlook the importance of pathologic diagnosis, even in situations where healthcare resources are scarce and clinical or radiological findings strongly indicate the presence of breast cancer. Advancements in artificial intelligence (AI) within the medical sector have opened up new possibilities for enhancing the precision of medical image analysis and decreasing the need for extensive human resources. AI excels in recognizing intricate patterns in images and quantifying data that may be challenging for humans to discern, thereby enhancing clinical decision-making processes [11] AI-based breast cancer imaging, pathology and adjuvant therapy technology cannot only reduce the workload of clinicians, but also continuously improve the accuracy and sensitivity of breast cancer diagnosis and treatment [12] Several past studies indicate that AI demonstrates satisfactory diagnostic accuracy in interpreting screening mammograms independently, akin to an individual reader. Present studies on AI in breast imaging primarily concentrate on detecting and distinguishing between benign and malignant lesions, forecasting molecular typing, evaluating risks, segmenting images, devising radiotherapy plans, and monitoring effectiveness. Nevertheless, the influence of these AI technologies on categorizing benign and malignant breast lesions through dynamic contrast-enhanced magnetic resonance imaging (DCE-MRI) remains constrained, necessitating further exploration in this domain.

A. Traditional Diagnostic Methods

1. Mammography and Imaging Techniques

Mammography remains the cornerstone of breast cancer screening and diagnosis. It involves the use of low-dose X-rays to create detailed images of the breast, which can reveal the presence of tumors or abnormalities that may indicate cancer. Mammography is particularly effective at detecting calcifications and other subtle changes in breast tissue that could be early signs of cancer [13].

Advanced imaging techniques such as ultrasound and MRI are often used in conjunction with mammography to improve diagnostic accuracy. Ultrasound, which uses sound waves to produce images, is especially useful for distinguishing between solid masses and fluid-filled cysts and is often used to evaluate suspicious areas found during a mammogram [14]. MRI, or magnetic resonance imaging, provides highly detailed images of breast tissue using magnetic fields and radio waves, and it is particularly useful for screening women at high risk of breast cancer and for assessing the extent of the disease in newly diagnosed patients [15].

2. Biopsy and Pathological Analysis

When imaging techniques identify a suspicious area, a biopsy is usually performed to obtain a tissue sample for pathological analysis. This is the definitive method for diagnosing breast cancer. Several types of biopsies can be performed, including fine-needle aspiration (FNA), core needle biopsy, and surgical biopsy. Each method has its own indications based on the size, location, and characteristics of the lesion [16].

Pathological analysis involves examining the tissue sample under a microscope to identify cancer cells and determine the type, grade, and other characteristics of the tumor. This information is crucial for planning treatment. For instance, the presence of hormone receptors or HER2 protein on cancer cells can significantly influence treatment decisions [17].

B. AI Applications in Breast Imaging and Pathology

Artificial Intelligence (AI) is increasingly being integrated into breast imaging and pathology to enhance diagnostic accuracy and efficiency. AI algorithms, particularly those based on deep learning, have shown promise in various aspects of breast cancer diagnosis, from image analysis to risk prediction.

1. AI in Breast Imaging

AI applications in breast imaging include improving the interpretation of mammograms, ultrasound, and MRI images. AI algorithms can assist radiologists by highlighting areas of concern, thus reducing oversight and increasing the detection rates of early-stage cancers. For instance, deep learning models have been trained to analyze mammograms with accuracy comparable to experienced radiologists, identifying patterns and anomalies that might be missed by the human eye [18]. AI is also being used to enhance the specificity of imaging techniques, reducing false positives and unnecessary biopsies. For example, AI-enhanced ultrasound can better differentiate between benign and malignant lesions, while AI applications in MRI can improve the assessment of tumor margins and detect metastases more accurately [19].

2. AI in Pathological Analysis

In pathology, AI tools are being developed to automate the analysis of biopsy samples. These tools can assist pathologists by quickly identifying cancerous cells in tissue samples, measuring tumor characteristics, and even predicting outcomes based on histological patterns [20]. AI can also facilitate the quantification of tumor markers, which is essential for personalized medicine. For example, AI algorithms can evaluate the expression of hormone receptors and HER2 status more consistently and accurately than manual methods, leading to more precise treatment plans [21]. Overall, the integration of AI in breast cancer diagnosis holds the potential to significantly improve the accuracy and efficiency of both imaging and pathological analysis, leading to earlier detection and better patient outcomes.

III. AI in Breast Cancer Prognosis and Prediction

A. Genomic and Molecular Analysis

The integration of artificial intelligence (AI) into genomic and molecular analysis has revolutionized the prognosis and prediction of breast cancer. This approach leverages advanced machine learning algorithms to analyze complex genomic datasets, leading to significant improvements in the accuracy of breast cancer prognosis and prediction. One significant advancement is the use of machine learning-based epigenetic classifiers to predict axillary lymph node (ALN) involvement in breast cancer patients. These classifiers utilize DNA methylation (DNAm) profiles from primary tumor specimens to identify patients with axillary lymph node metastasis with high accuracy. In a study, machine learning approaches generated five epigenetic classifiers with higher discriminative potential (AUC > 0.88) compared to traditional clinicopathological variables [22]. Furthermore, genomic analysis through RNA sequencing (RNA-seq) has been employed to discriminate between early and late stages of invasive ductal carcinoma (IDC). Supervised machine learning algorithms and feature selection methods such as Recursive Feature Elimination (RFE), Regularized Least Absolute Shrinkage and Selection Operator (RLASSO), and Random Forest have been used to identify gene features that can efficiently classify tumors based on their stage-specific gene expression profiles [23]. This approach has shown promise in capturing the cryptic signatures inherent in large-scale genomic data, facilitating the identification of prognostic biomarkers and the development of personalized treatment strategies. Additionally, mechanistic models incorporating biological knowledge have been developed to describe metastatic dynamics. These models, combined with machine learning techniques, offer a personalized prediction tool for metastatic relapse. For instance, a mechanistic model based on two intrinsic mechanisms of metastatic progression—growth and dissemination—demonstrated similar predictive performance to random survival forests and Cox regression models [24]. This model provides insights into the metastatic burden at diagnosis and simulates future metastatic growth, aiding in the selection of appropriate adjuvant therapies. The integration of AI in genomic and molecular analysis extends to radiogenomics, which combines radiomic data with genetic information to create non-invasive biomarkers. This fusion aims to predict the risk and outcomes of breast cancer more accurately. For example, radiomic signatures derived from magnetic resonance imaging (MRI) have been shown to predict axillary lymph node metastasis and disease-free survival in early-stage breast cancer patients. These signatures, when combined with molecular subtype information, enhance the predictive power and offer a preoperative approach to guide clinical practice [25]. In conclusion, AI-driven genomic and molecular analyses have significantly advanced the prognosis and prediction of breast cancer. By integrating machine learning algorithms with genomic data, researchers can develop robust predictive models that improve disease stratification, facilitate personalized treatment strategies, and ultimately enhance patient outcomes.

Conclusion:

In conclusion, AI combined with molecular and genomic analysis has significantly improved breast cancer outcomes. AI tools assist in imaging and pathological analysis, enhancing prediction and management, leading to earlier detection and prompt intervention. Specialists using AI-driven interventions can make better decisions. In spite of deficiencies in traditional diagnostic methods of Breast Cancer, most physicians are still using these methods. Fortunately, rapid development of AI(Artificial Inteligence) has come to our aid.AI can serve in different aspects of breast cancer management including prediction and evaluating the risk of getting cancer ,early detection, and analysing both pathological data and imaging with more accuracy and sensitivity.

Future direction:

This paper is a systematic review which investigates the application of artificial intelligence to breast cancer screening and diagnosing tests. The result of this research demonstrated that AI is being progressively involved into pathology and breast imaging in order to boost diagnostic efficiency and accuracy, which will result in earlier identification and improved outcomes for patients. Furthermore, the prognosis and prediction of breast cancer have greatly improved as a result of AI-driven genomic and molecular analysis. Researchers can create reliable predictive models that boost patient outcomes by facilitating individualized treatment options, improving illness stratification, and merging machine learning algorithms with genomic data. Suzanne C.Wetstein et al. 2020 created and verified an automated technique for breast terminal duct lobular unit (TDLU) involution measurement in order to deep learning assessment of TDLU involution , which serves as a preliminary step towards automated risk prediction for breast cancer which can be used more widely to estimate the risk of breast cancer in epidemiological research [26]. Javier I.J Orozco et al. 2022 found that machine learning -based epigenetic classifiers for axillary staging of patients with ER-positive early-stage breast cancer ma effectively separate patients with axillary disease from those who do not have lymph node involvement. Finding a reliable, non-invasive, facile to use technique for axilla staging can give the necessary prognostic information without the risks associated with surgery [22]. There were certain restrictions on or investigation. The most important one is the comparatively small number of examples in the context of AI effect on breast cancer, which is covered throughout the paper. More cases from various universities are required to enhance and refine our finding for possible application of AI in a therapeutic context. In other words, to ensure clinical effectiveness and generalizability, medical organization, AI developers, researchers, and governors must collaborate to assist make developing population-based imaging datasets from diverse communities available for external evaluation. Furthermore, we think that additional cancer types for which there are comparable concern regarding their growing worldwide occurrence could benefit from AI and thus this why an international multicenter study with an adequate number of sample size must be caried out in the future.

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