

Breakthroughs in Artificial Intelligence in Breast Cancer Diagnosis and Prognosis: Radiomics and Pathology

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Abstract. As the most common cancer after lung cancer presently, breast cancer needs to gain extensive awareness of both general public and healthcare professionals. With continuous developments of technology researchers on medical science began to attach great significance to the integration of artificial intelligence (AI) and clinical diagnostics. With the purpose of enhancing early diagnosis and prognosis and implementing more effective and timely treatment for breast cancer, increased research efforts are currently aimed at applying AI to breast cancer detection and classification, opening up new avenues for improving patient care. The paper initially discusses the progress of AI in the diagnosis and prognosis of breast cancer and presents an overview of the existing literature of research in this area. It describes the main methods and procedures employed in AI-assisted diagnosis and response prediction with particular emphasis on radiological as well as pathological approaches. Comparative analysis is also provided comparing differences in sensitivity and specificity between clinicians and AI systems when interpreting images. Furthermore, the paper discusses existing challenges in the introduction of AI in clinical practice to assist clinicians' diagnosis and carry out treatment. The major challenges include requirements for data volumes, standardization and ethical problems. Then, it predicted the future progress of AI technologies in diagnostics and prognosis of breast cancer, providing practical suggestions on promoting applications of AI in diagnosis, therapy and general use.

Keywords: Breast cancer, AI, Deep learning, Pathology, Imaging.

1. Introduction

Breast cancer, considered the most dangerous type of cancers for women, is caused by factors like genetic factor, hormone and environments. It holds the highest incidence rate and mortality in women, possibly triggered by malignant transformation of mammary epithelial cells. According to the global cancer statistics 2022 published by the International Agency for Research on Cancer (IARC) in 2024, breast cancer is the most common type except lung cancer [1]. The proportion is 11.6% of patients suffering from cancers globally. Meanwhile, breast cancer ranks fourth among cancer-related deaths, accounting for 6.9% of all death cases related to cancer. Current diagnostic methods of breast cancer are mainly ultrasound, mammography, the core biopsy, and lymph node biopsy. However, owing to diverse obstacles like variations in clinical expertise and delayed medical diagnosis, patients may lose opportunities to get in time treatment [2]. For those reasons, high recurrence rate and inhibition effect for therapies are also possible to emerge. Although researchers persistently attempt to apply technologies like genetic engineering and immunotherapy to clinical treatment, the outcomes still depend on the initial diagnostic results. Only the accurate and timely diagnosis can create a solid foundation for applications of advanced medical therapy. Apparently, there remains an extensive urgent demand for clinically accurate early detection of breast cancer.

With continuous development of technology, researchers have attempted to apply the advanced artificial intelligence (AI) technologies in the diagnostic of breast cancer. At present, the dominant trend is the introduction of deep learning algorithms in image analysis and classification of breast cancer. For its abilities to increase the accuracy of diagnostic detection in breast cancer, AI is capable of helping radiologists to make reliable decisions. Among the deep learning models, convolutional neural networks (CNNs) have attracted enormous attention. Compared with other models, deep CNN is the major algorithm for computer vision tasks. This characteristic possibly implies the potential of

CNN to become one of the most promising AI technologies in future applications [3]. For example, Masud et al. proposed a shallow custom CNN model and investigated its ability to classify breast cancer based on ultrasound images [4]. In comparison with other models, it is reported that the shallow CNN model showed a faster training speed and its accuracy of classification can be as high as 100%. It is a breakthrough of vital theoretical and practical basis for utilizing CNN models precisely diagnose breast cancer later. Furthermore, Desai et al. compared a multi-layer perceptron neural network (MLP) with a CNN and noted that, although the two models have similar image processing speeds, the CNN was superior to the MLP in terms of diagnostic accuracy in breast cancer detection [5]. Their research revealed the superiority of CNN in clinical diagnosis and offered interesting insights for future investigations.

In breast cancer prognosis, biomarkers are the first choice of disease assessment. Nevertheless, most conventional biomarkers have low sensitivity for low-volume diseases and are not perfect for screening or initial diagnosis. Recent studies show that small extracellular vesicle (sEV) proteins may be potential biomarkers with outstanding prognostic value. For instance, Chaudhary et al. reported that serum exosomal-annexin A2 is apparently overexpressed in triple-negative breast cancer (TNBC) patients. Its expression levels are also associated with tumor grade, representing a direct proportion [6]. The findings indicate that the protein is able to distinguish between breast cancer patients and healthy individuals and be utilized as a prognostic marker for patient prognosis. Nonetheless, despite these attempts, there remain some methodological challenges: current isolation techniques struggle to achieve both high yield and high purity of sEV proteins, the separation process is lengthy and labor-intensive, and contamination risk to the sample remains high.

Inspired by AI, some researchers developed a machine learning model, Mirai, for breast cancer risk prediction among originally cancer-free patients [7]. With the aid of this study, Damiani et al. conducted the research using the Mirai model to forecast the stage and grade of breast cancer [8]. After the process, they carried out a long-term follow-up for enrolled patients for at least three years. Their analysis revealed that the model had excellent predictive performance, especially in detecting high-grade breast cancers.

Though a great number of studies have emphasized the possibilities of AI for the diagnosis, grading, and prognostic evaluation of breast cancer, few challenges remain in their full integration into clinical practice. They are the fact that it is still unable detect the presence of cancer with 100% accuracy and ethical and moral concerns, which are issues that still have to be researched more [9]. Given these limitations and rising uses of AI-aided image analysis and clinical decision support, this study aims to sum up current research and propose practical recommendations to apply AI in breast cancer management. This attempt is hoped to pave the way and provide new inspirations for ongoing research in this field.

2. Methodology

2.1. Radiomics

In radiomics, AI is dominated by the application of Deep Learning. It is applied majorly in three fields, which include mammography (MMG), ultrasound (US), and breast magnetic resonance imaging (MRI) respectively. Mammography encompasses both digital mammography (DM) and digital breast tomosynthesis (DBT). DM provides conventional two-dimensional images, while DBT is an advanced pseudo-three-dimensional technology that is derived from DM with better cancer detection rates [10]. In digital mammography, Romero-Martín et al. have suggested that using AI as an independent screening reader can potentially reduce the workload of radiologists by up to 100% [11]. Meanwhile, recall rates can be reduced and unnecessary biopsies can also be avoided in this way. Contrary to this, AI systems applied in DBT have the tendency to produce higher rates of recall. However, these labeled cases can be reviewed again by radiologists as part of a second review, thus increasing overall detection efficiency and diagnostic accuracy.

Compared with its applications in MMG and MRI, studies of AI in breast ultrasound are quite rare. However, this does not diminish its clinical importance. In a research by Zhou et al., three types of CNN models, ResNet-101, Inception-ResNet V2, and Inception V3, were trained and tested by utilizing deep learning methods [12]. After making a comparison with the performance of some radiologists, these models showed capacity for predicting clinically negative axillary lymph node metastasis from primary breast cancer patients' ultrasound images. To its credit, Inception V3, which is the best-performing model, reached a sensitivity of 85% and a specificity of 73%, higher than that of the human readers, 73% and 63%, respectively. These results indicate the enormous potential of AI in clinical diagnosis and decision-making for breast ultrasound.

In order to determine whether AI can facilitate radiologists to interpret breast MRI and improve diagnostic efficacy, Jiang et al. conducted a study with radiologists from diverse clinical backgrounds and let them review dynamic contrast material-enhanced breast MRI (DCE-MRI) images [13]. Researchers constructed an AI system which is able to segment DCE-MRI images automatically and extract features. This study indicated that radiologists had stronger abilities to differentiate malignant lesions from benign lesions with the assistance of AI. To be specific, there was an increase from 0.71 to 0.76 in the average area under curve (AUC) values of clinicians after introducing AI to help interpretation. This kind of change means that the accuracy of diagnosis has been promoted, and the results are also more credible. However, the system provided minimal benefit in detecting whether patients are ill or not.

In terms of prognosis, AI can also leverage its considerable advantages within radiomics. Primary systemic therapy (PST), recognized for its ability to downstage tumors and minimize the extent of surgery, has been widely adopted by patients with locally advanced breast cancer [14]. Researchers have proposed that integrating AI into MRI—the preferred approach for assessing treatment response to PST—could make use of the strength of AI to enhance image analysis. In that case, prognostic evaluation for patients undergoing PST could be improved.

2.2. Pathology

In pathological diagnosis and practice, AI provides support to pathologists by analyzing the whole slide imaging (WSI) and hence aid breast cancer diagnosis and grading. In research by Mantrala et al., a deep learning algorithm model following the Nottingham Grading System (NGS) was applied and used to evaluate a scanned slide of 137 invasive carcinomas. The results then were compared to those of six pathologists [15]. This study revealed that judgements of AI are quite similar to decisions of pathologists when it comes to histologic grading. The result implied important information for further study to apply AI into diagnosis and grading accuracy in cancer pathology.

In clinical pathology, AI can also predict the therapeutic responses of breast cancer patients and thereby assist doctors in prognostic assessment and timely adjustment of treatment protocols. In order to counteract the risk of overtreatment in some patient subsets, Aswolinskiy et al. developed an explainable computational biomarker that would be able to forecast the responses to treatment before neoadjuvant chemotherapy (NAC) begins and thereby mark those patients who have the highest potential to benefit from this treatment [16]. Receiver operating characteristic (ROC) curves indicated that the biomarker achieved outstanding sensitivity, even up to 100% in triple-negative breast cancers (TNBC) and Luminal B breast cancer, implying that it is of high ability to estimate whether patients would respond to NAC therapy. The results suggest strong capacity of this biomarker for prognostic application and clinical application. In addition, Challa et al. proposed a new AI algorithm to identify lymph node (LN) metastasis of invasive breast cancer from screening hematoxylin and eosin (H&E)-stained slides [17]. Consistent with the expectation, it truly decreases the time spent assessing LN metastasis, enhances the efficiency of pathologists, and potentially can help improve patients' prognosis. Compared to conventional clinical scores, AI may assume a more objective role in slides evaluation, resulting in improved diagnostic reliability and accuracy.

However, Jarkman et al. have described significant limitations of pre-trained deep systems for detecting breast cancer metastasis in sentinel lymph nodes [18]. With multi-center data, the authors

concluded that model performance was distinctly impacted by changes in indications. Small variations resulted in significant reductions in predictive performance so that frequent retraining is necessary to obtain reliable models. These results emphasize that in order to develop clinically mature AI models for pathological prognosis of breast cancer, substantial challenges in model generalizability still need to be overcome.

Overall, the literature covers a broad spectrum of research on AI applications in pathology and radiology with useful methodological lessons and results for future studies. There is, however, a common feature in a lot of literature that is the general use of the AUC as the only, or at least main, performance indicator for evaluating the performance of AI-based diagnosis systems. Better AUC values tend to be implied better model performance and diagnostic accuracy. The high values that have been reported in most studies after AI applications are seen to approximate 1. This similarity shows the effectiveness and reliability of their experimental settings and models. In the meanwhile, the practical potential of AI in medical diagnosis is also indirectly revealed.

3. Challenges and Future Perspectives

Though plenty of researchers have explored using AI to promote diagnostic and prognostic advancement of breast cancer, this immature technology still has some defects. It is hindered by the requirement of large amounts of data because only extensive data sets can give adequate analytical performance [19]. On this basis, data acquisition and collection from various sources has become a fairly major problem, and effective data management has also become a pressing concern for researchers. In addition to technical constraints, the clinical application of AI also raises very serious ethical concerns. For instance, when AI provides erroneous diagnosis leading to adverse consequences, who is responsible for such medical accidents is also one of the issues that require resolution during the next phase. Moreover, one practical barrier still exists in the form of limited research funding [20]. Despite the support funds channeled by institutions such as the National Natural Science Foundation of China (NSFC), technological levels of development or regions have created unequal distribution of resources, thus restricting the popularization of funding project.

To overcome such barriers and further advance AI-assisted diagnosis and prognosis in breast cancer, future research needs to aim towards developing open-access databases and sharing common models [21]. More importantly, patients' privacy should be taken into consideration since data security is also a significant issue for AI. Governments out to respond to the development of technologies by formulating laws in order to guide hospitals and medical institutions. It is crucial for governments to determine the distribution of responsibilities when AI is engaged in detection and prediction of breast cancer. Such complete regulations can minimize the potential risks of AI-related accidents and offer guiding significance to clinicians. Equally, aiming at diminishing the restrictions of funds to technical development, sustained and reasonable investment is supposed to be guaranteed.

In order to reduce algorithmic bias and discrimination caused by sample data, more efforts should be put into maintaining trust from patients to medical professionals and ensuring fairness of AI. Meanwhile, it is of great importance to achieve algorithmic transparency [22]. With the persistent development of advanced technology and clinical application, it is necessary for clinicians to receive intensive training. As providers of medical service, medical workers must be equipped with professional skills and essential ethical knowledge. This kind of awareness drives clinicians better evaluate the possible ethical risks and promote harmonious doctor-patient relationships [23]. Furthermore, radiologists and pathologists specifically should be familiar with the utilization of AI systems and receive comprehensive training in AI-assisted cancer detection, classification and prognosis. As the integrating AI into medical care is a hot topic of investigation in the 21st century, medical practitioners and institutions are expected to keep aware of remarkable ethical consciousness and expertise. On this basis, addressing clinical challenges and medical problems for patients by taking advantage of AI would be their aim and duty.

4. Conclusion

Recent research has fully reflected the practical potential of AI in detection and prognosis of breast cancer. Those achievements also indicate diverse directions for clinical applications of AI. With the assistance of AI, recall rates are clinically reduced and some unnecessary biopsies can also be omitted. In this case, it is beneficial to improve detection efficiency and avoid delays in diagnosis. In addition, benefiting from support of AI, clinicians are able to identify benign and malignant MRI lesions more accurately, raising diagnostic accuracy. Moreover, in prognosis, AI is able to predict patients' response to current therapy and assess lymph node metastasis. Owing to this characteristic, medical workers are allowed to formulate appropriate therapeutic schedules based on individuality. However, there still remain a few challenges of clinical application. Enormous data requirements for analysis, limited data resources, responsibility assignment and financial problems are the prominent obstacles. Nonetheless, applying AI to medical diagnosis is an inevitable trend. Though obstacles mentioned above demand timely and effective solutions before the general applications, there still remains a promising prospect for AI. Future developments of AI technology show fundamental guiding significance to the diagnostics of breast cancer, finally optimizing the efficiency and accuracy of medical detection.

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