World Journal of Advanced Research and Reviews
eISSN: 2581-9615 CODEN (USA): WJARAI
Cross Ref DOI: 10.30574/wjarr
Journal homepage: https://wjarr.com/

(Review Article)

Check for updates

Deep learning in medical imaging for disease diagnosis

Mohammad Mojtaba Rohani [1,*], Seyedhassan Sharifi [2] and Soheil Durson [3]

[1] *Department of Radiology, Poursina Hospital, Guilan University of Medical Sciences, Rasht, Iran.*
[2] *Pediatric Cardiology Subspecialist, Pediatric Department, Day General Hospital, Iran.*
[3] *Information Technology Department, Izmir Yuksek Teknoloji Enstitusu, Türkiye.*

World Journal of Advanced Research and Reviews, 2025, 25(02), 2522-2526

Publication history: Received on 10 January 2025; revised on 22 February 2025; accepted on 25 February 2025

Article DOI: https://doi.org/10.30574/wjarr.2025.25.2.0558

Abstract

Deep learning plays a significant role in transforming medical imaging for disease diagnosis. It uses advanced algorithms, especially Convolutional Neural Networks (CNNs), to automatically learn and extract important features from medical images. This technology helps in detecting, classifying, and diagnosing various diseases, such as different types of cancer, brain disorders like aneurysms and strokes, heart diseases, and respiratory conditions. Deep learning improves the accuracy and efficiency of diagnostic workflows and reduces the workload for healthcare professionals. Despite its many advantages, deep learning faces challenges related to data availability, model interpretability, and clinical validation. This review highlights the current applications, performance evaluation methods, and challenges of deep learning in medical imaging for disease diagnosis.

Keywords: Artificial Intelligence; Deep Learning; Medical Imaging; Disease Diagnosis

1. Introduction

Medical imaging plays a crucial role in disease diagnosis and treatment planning by enabling non-invasive visualization of internal structures. Technologies such as X-ray, computed tomography (CT), magnetic resonance imaging (MRI), and ultrasound have revolutionized healthcare by providing critical insights into anatomical and functional abnormalities. These imaging modalities allow for the early detection of diseases, monitoring of disease progression, assessment of treatment efficacy, and guidance during surgical and interventional procedures [1-4]. For instance, MRI offers detailed soft -tissue contrast, making it invaluable in neurological and musculoskeletal imaging, while CT provides rapid, high-resolution cross-sectional images critical in emergency and trauma cases. Ultrasound, being portable and radiation-free, is extensively used in obstetrics, cardiology, and point-of-care diagnostics. Despite these advancements, interpreting medical images requires a high level of expertise and can be subject to inter-observer variability, where different radiologists may provide differing interpretations of the same image. This variability can lead to diagnostic errors, delayed treatment, and increased healthcare costs. Additionally, the growing volume of medical imaging data has placed an increasing workload on radiologists, leading to potential fatigue and burnout, which may further impact diagnostic accuracy. These challenges highlight the need for advanced computational tools to support and enhance the diagnostic process, paving the way for the integration of artificial intelligence and deep learning technologies in medical imaging.

Artificial Intelligence (AI) has emerged as a transformative force in medical applications, particularly through deep learning techniques. Deep learning, a subset of AI, utilizes neural networks to automatically learn hierarchical features from data, making it highly suitable for image-based tasks. Unlike traditional machine learning algorithms that rely on handcrafted features and domain-specific expertise [5,6], deep learning models can autonomously extract and optimize complex patterns from large datasets, significantly enhancing performance in tasks such as image classification, object detection, and segmentation [9,10,12]. AI has demonstrated remarkable potential in disease detection, classification,

* Corresponding author: Mohammad Mojtaba Rohani

and prognosis by automating and augmenting medical image interpretation [7,8,11]. Its applications span a wide range of clinical domains, including radiology, pathology, dermatology, ophthalmology, and cardiology. Additionally, AI-driven tools can assist in predicting disease progression, assessing treatment response, and stratifying patient risk, contributing to more personalized and precise medical care [13.14]. Among AI applications in healthcare, deep learning in medical imaging has gained significant traction. Convolutional Neural Networks (CNNs), a class of deep learning models, have shown state-of-the-art performance in analysing medical images for disease diagnosis [7,9,11]. CNNs are designed to automatically learn spatial hierarchies of features through layers of convolutional filters, pooling operations, and non-linear activations, enabling them to effectively capture intricate patterns within medical images. By leveraging vast amounts of imaging data, CNNs can assist clinicians in early diagnosis, improving accuracy and efficiency in medical decision-making. One of the key strengths of CNNs lies in their ability to generalize across diverse imaging modalities, including X-rays, CT scans, MRIs, ultrasound, and histopathological images. They excel in a variety of diagnostic tasks, such as detecting tumors, identifying fractures, segmenting organs, and classifying pathological conditions. For instance, CNN-based models have demonstrated remarkable performance in detecting breast cancer from mammograms, identifying pneumonia in chest X-rays, and diagnosing brain tumors from MRI scans, often achieving diagnostic accuracy comparable to or exceeding that of experienced radiologists.

2. CNN-Based Deep Learning in Disease Diagnosis

Convolutional Neural Networks (CNNs) have become the backbone of deep learning applications in medical imaging, excelling in tasks such as image classification, segmentation, and anomaly detection. These models can automatically extract spatial and hierarchical features from medical images, making them well-suited for diagnostic tasks. Unlike traditional image analysis methods that rely heavily on handcrafted features and domain-specific knowledge, CNNs learn relevant features directly from raw data through a series of convolutional layers, which enhances their adaptability and accuracy across various medical imaging modalities. CNN-based deep learning models have been successfully applied in detecting conditions such as lung cancer in CT scans, diabetic retinopathy in fundus images, and Alzheimer's disease in MRI scans. In lung cancer detection, for example, CNNs can identify subtle nodules or lesions in CT images that may be challenging for radiologists to detect, especially in early stages when the disease is most treatable. These models not only classify the presence of cancer but can also assist in quantifying tumor size, growth rate, and potential metastasis, which are critical for treatment planning.

Some of the CNN-based deep learning models have been more popular than others. ResNet, addresses the vanishing gradient problem in deep networks by utilizing residual learning. The architecture incorporates skip connections that allow gradients to flow through layers more effectively, enabling the training of very deep networks. ResNet has been widely used in medical imaging for disease classification, including cancer detection, pneumonia classification in chest X-rays, and brain tumor identification in MRI scans. VGG is a deep CNN architecture known for its simplicity and uniform design, with small 3x3 convolutional filters stacked sequentially. Despite its depth, VGG is computationally expensive compared to newer architectures. However, it has been successfully employed in various medical imaging applications, such as classifying skin lesions, diagnosing tuberculosis from X-ray images, and analyzing histopathological slides for cancer detection. The Inception architecture, developed by Google, utilizes multi-scale convolutional filters within a single layer, allowing the model to capture features at different scales efficiently. Its ability to balance accuracy and computational efficiency makes it suitable for medical imaging tasks, including retinal disease classification, brain hemorrhage detection, and mammographic mass classification.

Beyond these applications, CNNs are also employed in a wide range of diagnostic tasks, including the detection of pneumonia in chest X-rays, classification of skin lesions in dermatology [10], brain neurovascular diseases [9], and identification of cardiovascular diseases [7]. In pathology, digital histopathological slides are analyzed using CNNs to detect cancerous cells, grade tumors, and predict genetic mutations from tissue morphology. The versatility of CNNs extends to multi-modal imaging analysis, where data from different imaging techniques are combined to improve diagnostic accuracy. For instance, integrating PET and MRI data using CNN architectures can enhance the detection of brain tumors by leveraging both metabolic and anatomical information. Moreover, CNNs have facilitated advancements in real-time diagnostic support, particularly in emergency and critical care settings. AI-powered diagnostic tools can rapidly analyse medical images at the point of care, assisting clinicians in making timely decisions for conditions such as stroke, traumatic injuries, and acute coronary syndromes.

Despite their widespread success, the performance of CNN-based models depends heavily on the quality and diversity of the training data. Efforts are ongoing to address challenges such as data scarcity, class imbalance, and the need for explainable AI, which are essential for the safe and effective integration of CNNs into routine clinical workflows. Nonetheless, the continuous evolution of CNN architectures and training methodologies promises to further enhance their diagnostic capabilities and expand their applications in the future of medical imaging.

3. Applications of Deep Learning in Medical Imaging Across Various Diseases

Deep learning has profoundly impacted medical imaging, offering powerful tools for the detection, classification, and diagnosis of a wide range of diseases across multiple organ systems. One of the most significant applications is in the detection and characterization of tumors, where deep learning algorithms, particularly Convolutional Neural Networks (CNNs), have demonstrated exceptional performance in identifying malignant lesions in imaging modalities such as CT, MRI, PET, mammograms, and histopathological slides [15].

In neuroimaging, deep learning has been transformative in diagnosing and monitoring brain disorders, including brain tumors, aneurysms, strokes, Alzheimer's disease, and other neurodegenerative conditions [12]. For brain tumors, deep learning models can perform automated segmentation, detect tumor boundaries, and classify tumor types (e.g., gliomas, meningiomas) using MRI scans [15]. These models not only assist in initial diagnosis but also play a role in treatment planning by evaluating tumor progression and response to therapy. In the case of stroke, rapid diagnosis is critical for timely intervention. Deep learning algorithms can analyze non-contrast CT scans to detect early ischemic changes, intracranial hemorrhages, and large vessel occlusions with high accuracy, thereby expediting clinical decision-making in emergency settings. Additionally, in detecting cerebral aneurysms, deep learning models trained on vascular imaging modalities such as CT angiography (CTA) and magnetic resonance angiography (MRA) can identify aneurysmal dilations and predict the risk of rupture, supporting preventive care strategies [12]. Cardiovascular imaging has also greatly benefited from deep learning, particularly in the diagnosis of heart diseases such as coronary artery disease, heart failure, arrhythmias, and valvular disorders. Deep learning models can analyze echocardiograms, cardiac MRIs, and CT angiograms to assess cardiac function, detect structural abnormalities, and quantify parameters such as ejection fraction and myocardial strain [7]. For example, AI-powered tools can automatically identify coronary artery stenosis in CTA images, helping to stratify patients at risk for myocardial infarction. In arrhythmia detection, deep learning applied to electrocardiogram (ECG) data has shown impressive results in identifying irregular heart rhythms, such as atrial fibrillation, which may go unnoticed in standard diagnostic workflows [16]. Moreover, deep learning extends its applications to other critical areas of medical imaging, including the detection of musculoskeletal disorders, respiratory diseases, gastrointestinal conditions, and infectious diseases. In musculoskeletal imaging, deep learning algorithms assist in detecting fractures, joint abnormalities, and degenerative diseases from X-rays and MRIs. In respiratory diseases, such as COVID-19, pneumonia, and tuberculosis, deep learning models have been utilized to analyze chest X-rays and CT scans to identify infection-related patterns, assess disease severity, and monitor disease progression [17]. In summary, the applications of deep learning in medical imaging are vast and continuously expanding. These technologies not only enhance diagnostic accuracy and efficiency but also contribute to early disease detection, personalized treatment planning, and improved patient outcomes across a diverse range of medical conditions. As deep learning models continue to evolve with access to larger datasets and more sophisticated algorithms, their role in medical imaging will become increasingly integral to clinical practice, supporting healthcare professionals in delivering high-quality, data-driven care.

4. Current Challenges

Despite the success of deep learning in medical imaging, its integration into clinical practice is challenged by several interrelated issues. One primary concern is data availability and quality; obtaining large, diverse, and well-annotated medical imaging datasets is often restricted by privacy regulations, high costs of expert annotation, and other logistical hurdles, which in turn limits the amount of reliable data available for training robust models. This scarcity contributes to problems of generalization and bias, as models developed on narrowly defined datasets may not perform adequately when applied to broader or demographically diverse populations, potentially leading to skewed or inaccurate diagnostic outcomes. Additionally, the interpretability of these complex models remains a critical barrier; often operating as "black boxes," they provide little insight into their decision-making processes, thereby undermining clinician trust and making it difficult to validate or explain AI-driven conclusions. The computational costs associated with training such deep networks further complicate matters, as the need for high-performance computing resources can be prohibitive, particularly in resource-constrained settings. Finally, navigating the regulatory and ethical landscape is imperative, as any AI application in healthcare must adhere to strict standards designed to protect patient safety, ensure privacy, and maintain ethical integrity. Collectively, these challenges underscore the need for ongoing research and innovation to bridge the gap between the promising potential of deep learning and its practical, equitable, and transparent application in medical imaging.

5. Conclusion

In recent years, the field of medical imaging has been significantly transformed by the integration of deep learning techniques. Deep learning applications in disease diagnosis have been widely explored, and promising results have been demonstrated across various medical domains. Through the use of Convolutional Neural Networks (CNNs) and other advanced architectures, complex patterns in medical images have been effectively identified, and diagnostic processes have been greatly enhanced. Diseases such as cancer, brain disorders, cardiovascular conditions, and respiratory illnesses have been accurately detected, classified, and monitored with the support of deep-learning algorithms. The efficiency of diagnostic workflows has been improved, and the burden on healthcare professionals has been reduced through automation and decision-support tools powered by AI. Moreover, challenges related to inter-observer variability and human error have been mitigated, leading to more consistent and reliable diagnostic outcomes. Despite these advancements, several limitations must be acknowledged. Issues such as data scarcity, algorithmic bias, lack of interpretability, and the need for extensive clinical validation remain to be addressed. Additionally, ethical considerations and regulatory compliance must be carefully managed as AI-based systems are increasingly deployed in clinical practice. In conclusion, while significant progress has been made in applying deep learning to medical imaging, continuous research and development are required to overcome existing challenges. As new models are developed and larger, more diverse datasets are made available, the potential of deep learning to revolutionize medical diagnosis and improve patient care is expected to be fully realized.

Compliance with ethical standards

Disclosure of conflict of interest

No conflict of interest to be disclosed.

References

[1] Rahmani A, Norouzi F, Machado BL, Ghasemi F. Psychiatric Neurosurgery with Advanced Imaging and Deep Brain Stimulation Techniques. International Research in Medical and Health Sciences. 2024 Nov 1;7(5):63-74.

[2] Grignon B, Mainard L, Delion M, Hodez C, Oldrini G. Recent advances in medical imaging: anatomical and clinical applications. Surgical and Radiologic Anatomy. 2012 Oct; 34:675-86.

[3] Afrazeh F, Ghasemi Y, Shomalzadeh M, Rostamian S. The Role of Imaging Data from Different Radiologic Modalities During the Previous Global Pandemic. International Journal of Applied Data Science in Engineering and Health. 2024 Jul 27;1(1):9-17.

[4] Wagner Jr HN, Conti PS. Advances in medical imaging for cancer diagnosis and treatment. Cancer. 1991 Feb 15;67(S4):1121-8.

[5] Shruthi U, Nagaveni V, Raghavendra BK. A review on machine learning classification techniques for plant disease detection. In2019 5th International conference on advanced computing & communication systems (ICACCS) 2019 Mar 15 (pp. 281-284). IEEE.

[6] Norouzi F, Machado BL, Nematzadeh S. Schizophrenia Diagnosis and Prediction with Machine Learning Models. International Journal of Scientific and Applied Research (IJSAR), eISSN: 2583-0279. 2024 Dec 13;4(9):113-22.

[7] Sharifi S, Donyadadi A. Detection and Diagnosis of Congenital Heart Disease from Chest X-Rays with Deep Learning Models. International Journal of Applied Data Science in Engineering and Health. 2025 Jan 2;1(1):1-9.

[8] Abbasi H, Orouskhani M, Asgari S, Zadeh SS. Automatic brain ischemic stroke segmentation with deep learning: A review. Neuroscience Informatics. 2023 Sep 22:100145.

[9] Sharma P, Berwal YP, Ghai W. Performance analysis of deep learning CNN models for disease detection in plants using image segmentation. Information Processing in Agriculture. 2020 Dec 1;7(4):566-74.

[10] Chen H, Kim S, Hardie JM, Thirumalaraju P, Gharpure S, Rostamian S, Udayakumar S, Lei Q, Cho G, Kanakasabapathy MK, Shafiee H. Deep learning-assisted sensitive detection of fentanyl using a bubbling-microchip. Lab on a Chip. 2022;22(23):4531-40.

[11] Li H, Pan Y, Zhao J, Zhang L. Skin disease diagnosis with deep learning: A review. Neurocomputing. 2021 Nov 13; 464:364-93.

[12] Abbasi H, Afrazeh F, Ghasemi Y, Ghasemi F. A Shallow Review of Artificial Intelligence Applications in Brain Disease: Stroke, Alzheimer's, and Aneurysm. International Journal of Applied Data Science in Engineering and Health. 2024 Oct 5;1(2):32-43.

[13] Kukalakunta Y, Thunki P, Yellu RR. Deep Learning-Based Personalized Treatment Recommendations in Healthcare. Hong Kong Journal of AI and Medicine. 2024 May 1;4(1):30-9.

[14] Petrovic K. Deep Learning in Personalized Medicine: Advancements and Applications. Journal of Advanced Analytics in Healthcare Management. 2023 Mar 2;7(1):34-50.

[15] Aamir M, Rahman Z, Dayo ZA, Abro WA, Uddin MI, Khan I, Imran AS, Ali Z, Ishfaq M, Guan Y, Hu Z. A deep learning approach for brain tumor classification using MRI images. Computers and Electrical Engineering. 2022 Jul 1; 101:108105.

[16] Jamunadevi C, Bharanitharan J, Deepa S, Antony TJ. Brain Tumor Prediction from EEG Signal using Machine Learning Algorithm. In2023 4th International Conference on Electronics and Sustainable Communication Systems (ICESC) 2023 Jul 6 (pp. 1083-1089). IEEE.

[17] D'Angelo T, Caudo D, Blandino A, Albrecht MH, Vogl TJ, Gruenewald LD, Gaeta M, Yel I, Koch V, Martin SS, Lenga L. Artificial intelligence, machine learning and deep learning in musculoskeletal imaging: current applications. Journal of Clinical Ultrasound. 2022 Nov;50(9):1414-31