

Applications of Deep Learning in Medical Image Analysis: a Comprehensive Review of Recent Advances and Challenges

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ABSTRACT

Medical imaging serves as a cornerstone in modern healthcare, enabling non-invasive diagnosis, treatment planning, and patient follow-up through modalities like X-rays, CT scans, MRI, ultrasound, and PET scans. This comprehensive review delves into the transformative impact of deep learning on the field, mentioning core methodologies like CNNs, RNNs, Autoencoders, GANs, ViTs, and hybrid models. Applications are dissected across clinical domains including oncology, neurology, cardiology, pulmonology, ophthalmology, dermatology, and orthopedics, alongside tasks ranging from classification and segmentation to detection, registration, reconstruction, synthesis, and forecasting. Achievements are highlighted, particularly in achieving superior accuracy for classification and precise delineation in segmentation across varied medical contexts.

KEYWORDS

Deep learning; Medical image analysis; Convolutional neural networks; Explainable AI; Precision medicine

1 Introduction

Medical image analysis forms a fundamental pillar of contemporary healthcare. Interpreting these images, however, poses significant difficulties due to their complexity and patient-specific differences, often requiring expert radiologists and risking diagnostic variability or delays. As a subset of artificial intelligence, deep learning provides powerful solutions by leveraging extensive datasets and computational resources. Convolutional Neural Networks (CNNs), for example, excel in tasks such as classification, segmentation, detection, and alignment, sometimes surpassing human performance in focused applications. By extracting features autonomously from intricate patterns, these models accelerate and refine analytical processes, paving the way for tailored medical interventions. Nevertheless, substantial barriers persist, including data deficits, opacity in models, and challenges in adapting to varied populations and imaging protocols. These issues have prompted explorations of federated learning, explainable AI, synthetic data Preprint submitted to Elsevier creation, and ethical guidelines to promote reliability and equity.

This review delivers an exhaustive examination of deep learning’s role in medical image analysis, covering foundational technologies like CNNs and vision transformers, implementations in fields such as oncology and neurology, as well as enduring obstacles and emerging trajectories. Through systematic evaluation, it seeks to offer actionable insights for researchers and practitioners, thereby catalyzing continued progress in this evolving discipline.

2 Deep learning methodologies

Deep learning has fundamentally altered the landscape of medical image analysis, enhancing precision and operational speed in areas such as disease identification, anatomical delineation, and irregularity spotting by managing complex imaging datasets autonomously. Here, we examine the principal deep learning frameworks driving advancements in this area, with particular attention to their adaptations and optimizations suited to medical environments.

Table 1 Summary of classic CNN architectures in medical image analysis

Architecture	Key Innovation	Parameters (Approx.)	Medical Applications
AlexNet	ReLU, Dropout, Augmentation	60M	Breast cancer classification
VGGNet	Stacked 3x3 kernels	138M (VGG-16)	Pneumonia detection in X-rays
ResNet	Residual connections	25M (ResNet-50)	Lung cancer CT classification
U-Net5	Encoder-decoder with skips	31M	Brain tumor segmentation
DenseNet	Dense connectivity	8M (DenseNet-121)	Tuberculosis X-ray detection
EfficientNet	Compound scaling	5M (B0) to 66M (B7)	Lung/breast cancer imaging

Table 2 Other deep learning techniques in medical image analysis

Technique	Key Innovation	Main Applications
RNNs	Recurrent connections for temporal dependencies (e.g., LSTM, GRU)	Sequence analysis (e.g., fMRI, cardiac MRI, disease progression)
Autoencoders	Encoder-decoder for latent representation (e.g., VAE)	Denoising, reconstruction, anomaly detection
GANs	Adversarial generator-discriminator (e.g., CycleGAN, WGAN)	Data augmentation, cross-modal conversion, denoising
ViTs	Self-attention for global context (e.g., MedViT, Swin Transformer)	Classification, segmentation, detection of complex structures
Hybrid Models	Integration of architectures (e.g., CNN-LSTM, CNN-Transformer)	Complex tasks combining spatial/temporal/global features

2.1 Convolutional neural networks

Convolutional Neural Networks (CNNs) stand as a bedrock in medical image analysis, designed for grid-like data. Composed of convolution layers for spatial details, pooling to condense dimensions, and dense connections for predictions, these networks derive hierarchical attributes from pixels, bypassing handcrafted features and unraveling clinical visuals. In medical applications, CNNs are effective in tasks like categorization, delineation, localization, and alignment across formats such as radiographs, CT, MRI, and sonography, where certain implementations match or surpass specialist proficiency in delimited scenarios. Primary advantages include self-directed feature extraction, flexibility, and superior outcomes. Successive stages build representations from edges to forms, diminishing expertise dependence. They accommodate image quality fluctuations, artifacts, and standards, showing efficacy with collections like LIDC-IDRI or CBIS-DDSM^[1]. Table 1 encapsulates essential characteristics of seminal CNN designs.

2.2 Other deep learning techniques

Beyond Convolutional Neural Networks (CNNs), several other deep learning techniques have emerged as essential tools in medical image analysis. These include Recurrent Neural Networks (RNNs), Autoencoders, Generative Adversarial Networks (GANs), Vision Transformers (ViTs), Transfer Learning, and Hybrid Models. By leveraging mechanisms such as sequence dependency, unsupervised learning, adversarial training, global context modeling, and pre-trained architectures, these methods have advanced medical image processing in tasks like classification, segmentation, detection, denoising, and cross-modal conversion. Table 2 provides a comparative overview of these techniques, highlighting their key features, applications, advantages, challenges, and representative performance metrics.

3 Applications in medical imaging analysis

The application of deep learning in medical image analysis has significantly advanced the precision and automation of disease diagnosis, treatment planning, and condition monitoring across multiple domains, including oncology, neurology, cardiology, pulmonology, ophthalmology, dermatology, and orthopedics, as well as tasks such as classification, segmentation, detection, registration, reconstruction, synthesis, and prediction. This section systematically analyzes these applications through two perspectives: first, by medical domain, exploring the contributions of deep learning in specific clinical scenarios; second, by task type, examining the technological advancements along with their clinical impact.

3.1 Applications by medical domain

This part examines deep learning applications in oncology, neurology, cardiology, pulmonology, ophthalmology, dermatology, and orthopedics, encompassing classification, detection, segmentation, and prognostic prediction, with models, metrics, datasets, and emphasis on interdisciplinary collaboration.

3.1.1 Oncology

Oncology represents a primary domain for deep learning applications in medical image analysis, encompassing cancer classification, detection, segmentation, and prognostic prediction. EfficientNet-B7 achieved 99.14% accuracy in breast ultrasound classification, utilizing the BUSI dataset, with explainable AI techniques enhancing diagnostic transparency^[2]. A ResNet101-based CNN attained 96% accuracy in skin cancer detection on the ISIC-2018 dataset^[3]. In prognostic prediction, a ResNet-based model predicted TMB with AUC 0.971^[4]. Challenges include annotation costs and generalizability, suggesting data augmentation and transfer learning.

3.1.2 Neurology

In neurology, deep learning is employed for diagnosis and prediction of neurodegenerative diseases and epilepsy. Arafa et al. (2024) proposed a novel CNN architecture, achieving 99.98% accuracy on the Kaggle Alzheimer's dataset^[5]. Relative applications rely on multimodal data, with challenges in heterogeneity and consistency. Future research may explore multimodal fusion and self-supervised learning.

3.1.3 Cardiology

In cardiology, deep learning is utilized for segmentation, risk assessment, and detection. Kotte et al. (2025) employed hybrid 3D U-Net and attention mechanisms for whole-heart segmentation on CT images, outperforming methods on the MM-WHS dataset^[6]. For risk assessment, Revathi et al. (2024) developed an improved LSTM model, achieving 97.11% accuracy on the Cleveland dataset^[7]. While high-resolution imaging provides advantages for deep learning in cardiology, data privacy and model interpretability require further optimization.

3.1.4 Pulmonology

In pulmonology, deep learning targets diseases like tuberculosis and pneumonia. Mirugwe et al. (2025) utilized VGG16

to achieve 99.4% accuracy on chest X-ray images for tuberculosis classification ^[8]. Inbalatha et al. (2025) proposed EfficientNetB4 for high-accuracy detection in pediatric chest X-ray images ^[9]. Applications show potential for rapid diagnosis, with issues in data diversity and annotation quality.

3.1.5 Ophthalmology

In ophthalmology, DenseNet achieved 93.51% accuracy for early screening of diabetic retinopathy on the APTOS 2019 ^[10]. ResNet attained 99% accuracy for glaucoma on the ACRIMA ^[11]. U-Net combined with attention mechanisms achieved high Dice in retinal vessel segmentation ^[12]. These applications improve efficiency, though the detection of small lesions remains a challenge.

3.1.6 Dermatology

In dermatology, EfficientNetV2 achieved 96.04% accuracy for classification on the ISIC 2020 ^[13]. U-Net combined with attention mechanisms attained 0.937 DSC in skin lesion segmentation ^[14]. These applications have potential for remote diagnosis, but require ensuring generalizability across diverse skin tones.

3.1.7 Orthopedics

In orthopedics, Faster R-CNN achieved 66.82% accuracy for fracture screening on the MURA ^[15].

Table 3 Summary of applications in various medical fields

Medical Field	Task	Representative Model	Performance Metrics
Oncology	Breast Ultrasound Image Classification	EfficientNet-B7	Accuracy: 99.14%
Oncology	Skin Cancer Detection	ResNet101-based CNN	Accuracy: 96%
Oncology	Prognostic Prediction (TMB)	ResNet-based	AUC: 0.971
Neurology	Alzheimer's Disease Detection	CNN	Accuracy: 99.98%
Cardiology	Whole-Heart Segmentation	Hybrid 3D U-Net with Attention	Outperforms Traditional Methods
Cardiology	Cardiovascular Event Prediction	Improved LSTM	Accuracy: 97.11%
Pulmonology	Tuberculosis Classification	VGG16	Accuracy: 99.4%
Pulmonology	Pneumonia Detection	EfficientNetB4	High Accuracy
Ophthalmology	Diabetic Retinopathy Screening	DenseNet-based	Accuracy: 93.51%
Ophthalmology	Glaucoma Diagnosis	ResNet	Accuracy: 99%
Ophthalmology	Retinal Vessel Segmentation	U-Net with Attention	High Dice Coefficient
Dermatology	Skin Lesions Classification	EfficientNetV2	Accuracy: 96.04%
Dermatology	Skin Lesion Segmentation	U-Net with Attention	DSC: 0.937
Orthopedics	Fracture Screening	Faster R-CNN	Accuracy: 66.82%
Orthopedics	Tibiofemoral Joint Segmentation	U-Net	Dice: over 0.98 for certain bones

U-Net attained Dice over 0.98 for certain bones in tibiofemoral joint MRI segmentation ^[16]. These applications accelerate diagnostic processes, though refinement is required for complex patterns recognition.

3.2 Applications by task

This part comprehensively examines deep learning applications in classification, segmentation, detection, registration, reconstruction, synthesis, and prediction tasks. Each task is detailed with representative models, performance metrics, and datasets, underscoring its significance in clinical diagnosis and treatment while analyzing current challenges and future research directions.

3.2.1 Classification

Classification assigns labels to entire medical images, serving as a foundation for disease screening and diagnosis. Aftab et al. (2025) proposed a deep learning framework leveraging inverted residual and self-attention mechanisms, attained 98.6% accuracy in breast cancer classification on the IN-breast ^[17]. In this model, data augmentation addressed data imbalance and explainable AI improved diagnostic transparency. However, challenges persist, including class imbalance and cross-modal consistency. Future research may explore domain adaptation and multimodal data fusion to enhance model robustness and generalizability.

3.2.2 Segmentation

Segmentation partitions images into meaningful regions, forming a pivot of precise diagnosis and treatment planning. U-Net has become a seminal model for medical image segmentation, with its encoder-decoder structure and skip

connections. Nonetheless, challenges exist, including small targets and insufficient data for complex structures. Future research could leverage self-supervised learning, synthetic data generation, and federated learning to address data scarcity, with lightweight models to reduce costs.

3.2.3 Detection

Detection identifies specific objects or anomalies, which is critical for early screening. UrRehman et al.

(2024) proposed a CNN with dual attention, achieving approximately 95% sensitivity and 98% AUC on LUNA16^[18]. However, detecting small nodules and handling high-noise images remain challenging in low-dose CT scans. Future research may optimize multi-scale feature extraction and attention, with transfer learning for generalizability.

Table 4 Summary of tasks in medical imaging analysis

Task	Representative Application	Model	Performance Metrics
Classification	Breast Cancer Classification	IRCNN, SACNN	Accuracy: 98.6%
Segmentation	Medical Image Partitioning	U-Net	-
Detection	Lung Nodule Detection	Deep CNN with Dual Attention	Sensitivity: $\approx$ 95%, AUC: 98%
Registration	Brain MRI Registration	NIR	Faster optimization, low GPU memory
Reconstruction	Shoulder MRI Reconstruction	DL-MRI	Scan time reduced by 54.7%
Synthesis	MRI to CT Synthesis	CycleGAN	High image similarity, dose consistency
Prediction	Lung Disease Mortality Risk Prediction	CXR-Lung-Risk	C-index: 0.83

3.2.4 Registration

Registration aligns images from different sources or time to a unified coordinate system for multi-modal analysis and dynamic monitoring. NIR framework works model deformation fields, achieving performance comparable to traditional 3D brain MRI registration, with faster optimization and lower GPU memory usage^[19]. However, challenges include nonlinear mappings and costs for real-time applications. Future research may investigate hybrid models with attention mechanisms for efficiency and accuracy.

3.2.5 Reconstruction

Reconstruction aims to enhance image quality to reduce scanning time or radiation dosage. Liu et al. (2024) demonstrated DL-MRI reducing scan time by 54.7% in shoulder MRI while significantly improving quality and reducing artifacts^[20]. However, models require validation for generalizability across diverse devices and populations. Future research may explore transfer learning and domain adaptation for robustness in varied clinical settings.

3.2.6 Synthesis

Synthesis generates cross-modality images for analysis and reduced scans. Sherwani et al. (2024) reviewed deep learning applications in MRI, CT, PET image synthesis, encompassing architectures such as CNNs, Transformers, and diffusion models^[21]. Synthesis techniques reduce radiation exposure and costs. Nevertheless, training instability and the authenticity of generated images for high-resolution outputs remain challenges. Future research may explore advanced models and multimodal data fusion for quality and clinical applicability.

3.2.7 Prediction

Prediction forecasts disease progression or prognosis from imaging data. Weiss et al. (2023) developed the CXR-Lung-Risk model to predict lung disease mortality risk from chest X-ray images, achieving c-index 0.83 on the PLCO^[22]. The integration of 20 CNNs with multimodal data enhanced accuracy. Studies indicate that it supports personalized prevention and treatment strategies. However, adaptability and generalizability across racial groups require investigation. Future research may leverage federated learning and multimodal data fusion to bolster predictive capabilities in diverse populations.

4 Discussion on Challenges and Future Directions

Deep learning has profoundly advanced the precision and efficiency of diagnosis and treatment in medical image analysis. However, adoption faces challenges including data, model, and ethical issues. This section explores challenges, mitigation, impact on clinical translation, and propose future directions.

4.1 Data-related challenges

Deep learning models require large volumes of high-quality annotated data for training. However, medical datasets are smaller due to annotation costs and regulations like HIPAA and GDPR. This data scarcity can lead to overfitting, limiting performance on unseen data. For instance, high-quality annotated brain tumor MRI data is confined to specific projects, insufficient for large-scale model training^[23]. Researchers propose strategies like multi-task learning with shared

information, to address this challenge. However, authenticity and costs require optimization.

Medical imaging data exhibit diversity due to variations in devices, protocols, and populations, which often results in poor generalizability and compromising diagnostic accuracy. For example, a model trained on single-hospital CT performs poorly elsewhere^[24]. Domain generalization aims to train models for robustness without target data. Test datasets should encompass diversity, with initiatives collecting varied data. However, costs and protocols pose issues, in need of future harmonization and simulation.

4.2 Model-related challenges

Deep learning models are often perceived as ‘black boxes,’ a critical concern in medicine where insight into rationales is required to build trust. XAI enhances transparency by providing visual, textual, or example-based explanations^[25]. Effectiveness varies, with challenges in complexity and stability. Future research should develop standardized metrics and promote collaboration.

Deep learning models may amplify biases from training data, leading to suboptimal diagnostic performance for certain populations. Such biases exacerbate healthcare disparities, undermining model fairness. For example, models trained on racially homogeneous datasets may underperform on patients from other racial groups^[26].

Systematic reviews categorize fairness assessment and mitigation strategies like preprocessing, in-model debiasing, and post-processing. Balancing datasets reduces disparities. However, metrics and datasets need development, with future benchmarks and diverse designs.

4.3 Ethical and legal challenges

The application of deep learning in medical image analysis raises concerns in privacy, consent, and accountability. Privacy protection necessitates compliance with HIPAA and GDPR. Federated learning reduces and differential privacy risks. Transparent processes and auditing enhance compliance, advocating multidisciplinary guidelines. Legal frameworks delineate responsibilities, with surveillance for safety. Future research advances in technologies and harmonized standards, including case studies.

5 Conclusion

Deep learning has profoundly transformed medical image analysis, leveraging technologies such as Convolutional Neural Networks (CNNs) and Vision Transformers (ViTs) to achieve performance levels comparable to or surpassing human experts in tasks including classification, segmentation, detection, and registration. These advancements have significantly enhanced diagnostic accuracy and efficiency across diverse domains such as oncology, neurology, cardiology, pulmonology, ophthalmology, dermatology, and orthopedics, thereby advancing the paradigm of precision medicine.

Future research should prioritize addressing these challenges. Federated learning, by enabling distributed training, safeguards patient privacy. Explainable AI (XAI) methods, such as Grad-CAM, enhance model transparency by visualizing decision rationales. Multimodal data fusion, integrating imaging, genomic, and clinical data, holds promise for further improving diagnostic accuracy.

To fully realize the potential of deep learning, interdisciplinary collaboration is paramount. AI researchers, clinicians, ethicists, and policymakers must collaborate to establish standardized evaluation frameworks, fairness metrics, and ethical guidelines to ensure model robustness, transparency, and compliance. By developing lightweight models and real-time diagnostic tools, AI can be extended to resource-constrained settings, such as rural clinics, thereby promoting global health equity. In summary, deep learning is reshaping the future of medical image analysis, and through continued innovation and collaboration, it will significantly enhance patient care and healthcare system efficiency.

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