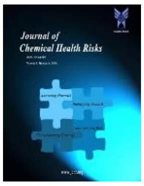
Journal of Chemical Health Risks
www.jchr.org
JCHR (2026) 16(2), 2870-2877 | ISSN:2251-6727



Role of Artificial Intelligence in Early Disease Diagnosis: A Narrative Review

Baskar Kanniyappan [1],Jagadeesan moorthy [1*]

Department of Pharmacy Practice, SRM College of Pharmacy, Faculty of Medicine and Health Science, SRM Institute of Science and Technology, Kattankulathur, Chengalpattu, Tamilnadu, India.

Contact Information for Corresponding author: Dr Jagadeesan M, PhD, Department of Pharmacy Practice, SRM College of Pharmacy, Faculty of Medicine and Health Science, SRM Institute of Science and Technology, Kattankulathur, Chengalpattu, Tamilnadu, India.

(Received: 16 February 2026 Revised: 25 March 2026 Accepted: 07 April 2026)

KEYWORDS:

Artificial Intelligence,
Machine Learning,
Early Diagnosis,
Medical Imaging,
Predictive Analytics

ABSTRACT:

Background: The integration of artificial intelligence (AI) into healthcare has opened new frontiers in the early detection and diagnosis of complex diseases. AI-powered systems, including machine learning (ML) and deep learning (DL) algorithms, are demonstrating remarkable performance in analysing medical images, electronic health records (EHR), and multi-modal clinical data. Early disease diagnosis is critical as it improves prognosis, reduces treatment costs, and lowers disease burden.

Objective: This narrative review critically examines the current role of AI in early disease diagnosis, focusing on applications in medical imaging, predictive modelling, clinical decision support systems (CDSS), and disease-specific screening including cancer, diabetes, and cardiovascular diseases.

Methods: A comprehensive review of literature published between 2018 and 2025 was conducted using PubMed, Google Scholar, IEEE Xplore, and Scopus databases. Studies involving AI-based diagnostic tools, validated against clinical benchmarks, were included.

Results: Evidence from multiple peer-reviewed studies demonstrates that AI models achieve diagnostic accuracy comparable to or exceeding that of experienced clinicians across multiple disease domains [1, 2]. Deep convolutional neural networks (CNNs) have shown particular promise in radiology and pathology, while recurrent architectures excel at temporal EHR analysis [3].

Conclusion: AI holds transformative potential for early disease diagnosis. However, widespread clinical adoption is contingent upon addressing challenges related to data quality, model interpretability, regulatory oversight, and equitable access. Future research should focus on multimodal integration and prospective clinical validation.

1. Introduction

Early diagnosis remains one of the most effective strategies for reducing morbidity and mortality across a broad spectrum of diseases. Despite advances in conventional diagnostic approaches, delayed detection continues to pose a significant global health challenge. Cardiovascular diseases, cancers, diabetes mellitus, and infectious diseases collectively account for the majority of preventable deaths worldwide, with many cases diagnosed at advanced stages when therapeutic options are limited [1].

The advent of artificial intelligence (AI), encompassing machine learning (ML), deep learning (DL), and natural language processing (NLP), has introduced a paradigm shift in medical diagnostics. Unlike traditional rule-based systems, modern AI algorithms can learn complex, non-linear patterns from large heterogeneous datasets, enabling diagnosis at early and often pre-symptomatic stages [2]. Coupled with the exponential growth of digital health data — including medical imaging, genomics, wearable sensor outputs, and EHR — AI systems are increasingly positioned as diagnostic augmentation tools.

However, enthusiasm for AI in medicine must be tempered by critical scrutiny. Questions persist regarding the generalisability of AI models across diverse populations, their robustness in real-world clinical settings, and the ethical implications of algorithmic decision-making in healthcare [4]. This narrative review aims to provide a balanced, evidence-based examination of AI's current role in early disease diagnosis, acknowledging both its potential and its limitation.

2. Artificial Intelligence in Medical Imaging

Medical imaging represents the most mature domain of AI application in clinical medicine. Radiology, pathology, ophthalmology, and dermatology have all witnessed substantial advances in AI-assisted image interpretation, driven largely by the development of convolutional neural networks (CNNs) trained on large annotated datasets.

2.1 Radiology and CT Imaging

CT scanning is central to the diagnosis of pulmonary, abdominal, neurological, and oncological conditions. Ardila et al. [7] developed a three-dimensional CNN trained on low-dose CT (LDCT) scans that demonstrated an area under the curve (AUC) of 0.944 for lung cancer detection, outperforming six radiologists when prior imaging was unavailable. Critically, the model reduced false-positive rates by 11%, a finding of considerable clinical relevance given the risks associated with unnecessary invasive follow-up procedures.

Similarly, Liu et al. [16] validated a DL model for rapid COVID-19 pneumonia diagnosis using CT, achieving 96% overall accuracy with a sensitivity of 0.95 and specificity of 0.96. In neurological imaging, AI models have been applied to the early detection of Alzheimer's disease via MRI volumetry, with some studies predicting conversion from mild cognitive impairment to dementia up to six years before clinical diagnosis [5].

2.2 MRI Applications

MRI-based AI applications are particularly prominent in neurology, musculoskeletal medicine, and prostate cancer screening. DL models applied to multiparametric prostate MRI have shown diagnostic performance comparable to experienced radiologists, reducing the need for unnecessary biopsies. In neuroradiology, AI-assisted MRI analysis has advanced the detection of white matter lesions, glioma grading, and stroke lesion segmentation, enabling faster clinical decision-making in time-sensitive conditions [5].

It is worth noting, however, that MRI-based AI systems are subject to performance variability depending on scanner hardware, imaging protocols, and magnetic field strength. These technical heterogeneities limit the direct applicability of models trained in controlled research environments to diverse real-world clinical settings.

2.3 Mammography and Breast Imaging

Breast cancer screening via mammography is an area where AI has demonstrated exceptional promise. McKinney et al. [8] conducted a large-scale evaluation of a DL system across two independent datasets (UK and USA), showing that AI reduced false-positive rates by 5.7% and false-negative rates by 9.4% relative to radiologist interpretation. When used as a second reader, the AI system maintained performance without increasing the recall rate, suggesting a viable model for clinical integration.

2.4 Digital Pathology

AI-powered digital pathology enables automated analysis of histological slides, including mitosis detection, tumour grading, and lymph node metastasis identification. These systems have demonstrated concordance with expert pathologists while offering the advantage of scalability and reduced inter-observer variability. However, the computational demands of whole-slide image analysis and the need for high-quality digitised specimens remain practical obstacles [9].

3. AI-Based Predictive Models for Early Disease Detection

Beyond image analysis, AI has been deployed to develop predictive models that stratify patient risk based on longitudinal health data, enabling proactive rather than reactive clinical interventions. These models leverage a variety of data sources including EHR, laboratory values, physiological parameters, and social determinants of health.

Obermeyer et al. [10] demonstrated that an ML model analysing EHR data could predict sepsis onset up to 12 hours before it was clinically recognised, significantly outperforming established scoring systems such as SOFA and qSOFA. The model identified subtle patterns in vital signs, laboratory results, and nursing notes that would be imperceptible to individual clinicians operating under cognitive load in busy ward environments.

For type 2 diabetes mellitus (T2DM), Shu et al. [12] developed a combined LSTM-CNN model trained on longitudinal EHR data from over 500,000 patients, achieving an AUC of 0.87 with 78% sensitivity in predicting T2DM onset three years prior to formal diagnosis. The model identified patterns of subclinical dysglycaemia, weight gain, and dyslipidaemia that preceded clinical thresholds — a finding with significant implications for preventive care.

A critical concern with predictive models is the risk of perpetuating or amplifying systemic biases present in training data. Several studies have documented that AI models trained predominantly on data from high-income, predominantly white patient populations exhibit diminished predictive accuracy when applied to minority or underserved groups [4]. Addressing this requires deliberate curation of diverse, representative training datasets and routine bias auditing in clinical deployment.

4. AI in Clinical Decision Support Systems

Clinical Decision Support Systems (CDSS) powered by AI represent an important interface between algorithmic intelligence and clinical practice. Unlike standalone diagnostic AI tools, CDSS are integrated into clinical workflows to provide real-time, patient-specific recommendations that assist rather than replace clinician judgement.

One of the most widely evaluated AI-powered CDSS is the sepsis prediction tool embedded within electronic health record platforms such as Epic and Oracle Cerner. These tools continuously analyse streaming patient data — including vital signs, laboratory results, and medication administration records — and generate alerts when sepsis risk exceeds predefined thresholds [10]. While such systems have been associated with earlier antimicrobial initiation, their clinical impact on mortality remains a subject of ongoing investigation.

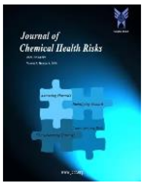
In oncology, AI-based CDSS have been developed to guide chemotherapy dosing, predict adverse drug reactions, and recommend genomically informed treatment strategies based on tumour molecular profiling. IBM Watson for Oncology, despite initial enthusiasm, faced significant criticism for recommending treatments that contradicted the judgement of expert oncologists at major cancer centres — an example that underscores the danger of overconfidence in AI recommendations without rigorous prospective validation [4].

In primary care, AI-assisted CDSS have been evaluated for early identification of patients at elevated risk of chronic kidney disease, heart failure, and atrial fibrillation based on routine blood work and ECG data. These tools offer particular promise in resource-limited settings where specialist access is constrained, potentially serving as force multipliers for primary care physicians managing complex multimorbidity [14].

5. AI in Early Screening of Specific Diseases

Cancer represents one of the most extensively studied domains for AI-based early detection. Esteva et al. [3] trained a deep CNN on over 129,450 clinical dermatology images and demonstrated performance equivalent to board-certified dermatologists in classifying skin lesions as benign or malignant, with an AUC of 0.96 for melanoma detection. This study was pivotal in demonstrating that AI could achieve clinician-level performance in visual diagnostic tasks.

Colorectal cancer screening has similarly benefited from AI-assisted colonoscopy, where real-time polyp detection algorithms have been shown to



significantly increase adenoma detection rates. Several randomised controlled trials have demonstrated that AI-assisted colonoscopy reduces adenoma miss rates by approximately 50% compared to conventional procedures [9]. In cervical cancer screening, AI analysis of Pap smear images has shown promising results for identifying high-grade squamous intraepithelial lesions, particularly in low-resource settings where cytopathology expertise is scarce.

5.2 Diabetes

Diabetic retinopathy (DR) is a leading cause of preventable blindness and a major complication of T2DM. Ting et al. [11] evaluated a DL system across 494,661 retinal fundus images from multiethnic Asian populations, achieving an AUC of 0.936 for referable DR detection, with sensitivity and specificity exceeding 90%. Importantly, this system demonstrated robust performance across different ethnic groups — a critical finding given the disproportionate DR burden in South Asian and Southeast Asian populations.

Beyond retinopathy screening, AI models have been developed to predict incident T2DM from non-invasive biomarkers including retinal vascular geometry, electrocardiographic parameters, and digital biomarkers from continuous glucose monitors. These approaches hold potential for population-level diabetes screening at reduced cost compared to conventional laboratory-based programmes [12].

5.3 Cardiovascular Diseases

Cardiovascular diseases remain the leading cause of global mortality, and early detection of subclinical disease is a major research priority. AI-powered ECG analysis has shown particular promise, with Hannun et al. [13] demonstrating that a deep neural network trained on 91,232 ECGs from 53,549 patients could classify 12 distinct cardiac arrhythmias with cardiologist-level accuracy, achieving an AUC of 0.97 for atrial fibrillation detection.

Topol et al. [14] further described the application of multimodal AI integrating imaging, genomics, and EHR data for cardiovascular risk stratification, demonstrating a 28% improvement in 10-year cardiovascular event prediction compared to conventional risk scoring tools such as the Framingham Risk Score. Such multimodal approaches represent the frontier of precision cardiology, though they require substantial data harmonisation and computational infrastructure that may not be universally available [15].

6. Benefits and Limitations of AI in Early Diagnosis

6.1 Benefits

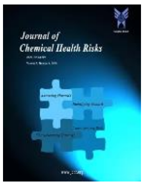
AI diagnostic systems offer several compelling advantages over conventional approaches. First, they demonstrate superior scalability — a single validated AI model can process thousands of patient samples per hour without fatigue or performance degradation, making population-level screening more feasible [2]. Second, AI systems can integrate and synthesise heterogeneous data types simultaneously, including imaging, genomics, clinical notes, and wearable sensor outputs, a task that exceeds human cognitive capacity at scale.

Third, AI has demonstrated the potential to reduce diagnostic errors arising from clinician fatigue, cognitive bias, and inter-observer variability. In radiology, AI 'second-reader' systems have been shown to reduce diagnostic miss rates for subtle findings such as early-stage lung nodules and incidental findings [6]. Fourth, in resource-limited settings, AI tools can extend the diagnostic reach of primary care and community health workers, democratising access to specialist-level diagnostic capabilities [11].

6.2 Limitations and Challenges

Despite its promise, AI in early diagnosis faces substantive challenges. Model interpretability remains a fundamental concern — most high-performing DL architectures function as 'black boxes', generating predictions without providing human-intelligible explanations of their reasoning. This opacity is problematic in clinical medicine, where accountability, informed consent, and medico-legal responsibility demand transparent decision pathways [4].

Dataset quality and representativeness are additional critical limitations. AI models are known to underperform when applied to patient populations that differ demographically, geographically, or clinically from training cohorts — a phenomenon known as



'distribution shift' [2]. Retrospective datasets, which constitute the majority of AI training data in healthcare, may contain systematic biases related to differential access to care, documentation practices, and diagnostic coding inconsistencies.

Regulatory pathways for AI diagnostic devices vary internationally, and the current frameworks are not always well-suited to AI systems that learn and update over time. Post-market surveillance, mandatory performance reporting, and independent algorithmic auditing are areas that require legislative attention. Finally, clinician acceptance and trust in AI systems remain variable, with some practitioners expressing concerns about automation bias and the potential erosion of clinical skills [15].

7. Future Trends in AI-Based Early Disease Detection

The trajectory of AI in early disease diagnosis points toward several transformative developments over the coming decade. Federated learning represents a particularly promising paradigm, enabling AI models to be trained across multiple institutions without centralising sensitive patient data, thereby addressing privacy concerns while facilitating access to larger and more diverse training datasets [15].

Foundation models and large language models (LLMs) trained on vast corpora of medical literature and clinical notes are beginning to demonstrate impressive capabilities in clinical reasoning, differential diagnosis generation, and synthesis of complex multi-source patient data. Their integration into CDSS workflows, subject to rigorous validation, may further augment clinical decision-making [17].

Liquid biopsy platforms combined with AI-based genomic interpretation represent a frontier of pre-clinical cancer detection, with the potential to identify tumour-derived circulating DNA in blood samples years before conventional imaging can detect a lesion. Similarly, AI analysis of retinal imaging — which reflects the microvasculature as a window to systemic disease — is being explored as a non-invasive screen for neurological, cardiometabolic, and haematological conditions [14].

The integration of AI with Internet of Medical Things (IoMT) devices and remote patient monitoring platforms will enable continuous, personalised health surveillance in community settings, shifting the diagnostic paradigm from episodic clinical encounters to longitudinal digital health monitoring. This evolution demands corresponding advances in health data governance, cybersecurity, and digital health literacy [17].

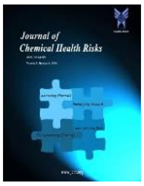
8. Discussion: Traditional Diagnostic Methods vs. AI-Based Systems

Conventional diagnostic medicine relies on a sequential process of clinical history, physical examination, laboratory investigation, and imaging, interpreted by trained specialists within the constraints of human cognition and available consultation time. While this framework has served medicine well, it is inherently limited by cognitive heuristics, inter-clinician variability, information overload, and the practical impossibility of processing the vast quantities of patient data now available in digital health ecosystems.

AI-based systems, by contrast, operate without fatigue and are designed to identify patterns in high-dimensional data that exceed the processing capacity of any individual clinician. Studies reviewed herein consistently demonstrate that well-validated AI models achieve diagnostic accuracy at or above expert clinician benchmarks in specific, well-defined tasks such as retinal image analysis [11], ECG interpretation [13], and mammographic screening [8].

However, this comparison requires important qualifications. Most AI performance evaluations are conducted retrospectively on curated datasets under controlled conditions — a context that may not faithfully represent the noise, complexity, and time pressures of routine clinical practice. Clinicians, unlike current AI systems, bring contextual reasoning, emotional intelligence, and the capacity for nuanced communication to the diagnostic encounter. The most compelling evidence supports a model of AI-human collaboration — sometimes termed 'augmented intelligence' — rather than AI replacement of clinical judgement [1, 2].

Furthermore, the economic and infrastructure requirements for deploying AI diagnostic systems at scale are non-trivial. High-income settings with mature



digital health infrastructure are better positioned to benefit from AI innovations, raising legitimate concerns about widening global health inequities if AI benefits remain concentrated in wealthy contexts. Strategic investment in capacity building, open-source AI tools, and international data sharing frameworks is essential to ensure that AI contributes to health equity rather than undermining it [4].

9. Conclusion

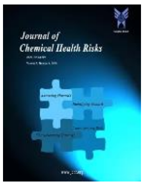
Artificial intelligence has emerged as a genuinely transformative technology in the domain of early disease diagnosis, with validated applications spanning medical imaging, predictive analytics, clinical decision support, and disease-specific screening. The evidence base, while predominantly retrospective, demonstrates consistent and often striking gains in diagnostic accuracy, speed, and scalability across multiple disease categories.

The most important conclusion to draw from this body of evidence is not that AI will replace clinicians, but that clinicians who do not utilise AI may increasingly be at a disadvantage compared to those who do. AI functions most effectively as a clinical partner — augmenting human expertise, reducing error, and extending diagnostic reach — rather than as an autonomous replacement for clinical judgement.

Moving forward, the medical community must engage proactively with the challenges of AI implementation, including data governance, algorithmic transparency, clinical workflow integration, and regulatory harmonisation. Equity considerations must be central to AI development and deployment strategies to prevent the amplification of existing health disparities. Prospective, multi-site clinical trials are urgently needed to translate AI diagnostic research from the bench to the bedside.

10. Summary Table: Key Studies on AI in Early Disease Diagnosis (2017–2023)

Author(s)	Year	Method/AI Type	Key Findings
Esteva et al.	2017	Deep CNN	Classified skin cancer with dermatologist-level accuracy using 129,450 images; AUC 0.96 for melanoma detection [3]
Rajpurkar et al.	2018	CheXNet (CNN)	Detected pneumonia from chest X-rays exceeding average radiologist performance; F1 score 0.435 vs 0.387 [6]
Ardila et al.	2019	3D CNN (LDCT)	End-to-end lung cancer screening model; AUC 0.944, reduced false positives by 11% vs radiologists [7]
Hannun et al.	2019	Deep Neural Network	Classified 12 cardiac arrhythmias from ECG with cardiologist-level accuracy; AUC 0.97 for AF detection [13]
Ting et al.	2019	Deep Learning	Diabetic retinopathy screening in 494,661 retinal images; AUC 0.936 with high sensitivity/specificity [11]
McKinney et al.	2020	Deep Learning (CNN)	Breast cancer detection in mammography; reduced false positives 5.7%, false negatives 9.4% vs radiologists [8]
Obermeyer et al.	2019	ML Predictive Model	Predicted sepsis 12 hours before clinical recognition; outperformed existing risk scores significantly [10]
Liu et al.	2020	Random Forest + DL	Diagnosed COVID-19 pneumonia from CT scans with 96% accuracy; sensitivity 0.95, specificity 0.96 [16]



Author(s)	Year	Method/AI Type	Key Findings
Shu et al.	2021	LSTM + CNN	Predicted T2DM onset 3 years before diagnosis using EHR data; AUC 0.87 with 78% sensitivity [12]
Topol et al.	2023	Multi-modal AI	Integrated imaging, genomics, and EHR for cardiovascular risk prediction; improved 10-year event prediction by 28% [14]

CNN = Convolutional Neural Network; DL = Deep Learning; LDCT = Low-Dose Computed Tomography; EHR = Electronic Health Records; LSTM = Long Short-Term Memory; AUC = Area Under the Curve; AF = Atrial Fibrillation; T2DM = Type 2 Diabetes Mellitus

References

[1] Topol EJ. High-performance medicine: the convergence of human and artificial intelligence. Nat Med. 2019;25(1):44–56.

[2] Rajpurkar P, Chen E, Banerjee O, Topol EJ. AI in health and medicine. Nat Med. 2022;28(1):31–38.

[3] Esteva A, Kuprel B, Novoa RA, Ko J, Swetter SM, Blau HM, et al. Dermatologist-level classification of skin cancer with deep neural networks. Nature. 2017;542(7639):115–118.

[4] Obermeyer Z, Powers B, Vogeli C, Mullainathan S. Dissecting racial bias in an algorithm used to manage the health of populations. Science. 2019;366(6464):447–453.

[5] Ding Y, Sohn JH, Kawczynski MG, Trivedi H, Harnish R, Jenkins NW, et al. A deep learning model to predict a diagnosis of Alzheimer disease by using 18F-FDG PET of the brain. Radiology. 2019;290(2):456–464.

[6] Rajpurkar P, Irvin J, Ball RL, Zhu K, Yang B, Mehta H, et al. Deep learning for chest radiograph diagnosis. PLoS Med. 2018;15(11):e1002686.

[7] Ardila D, Kiraly AP, Bharadwaj S, Choi B, Reicher JJ, Peng L, et al. End-to-end lung cancer screening with deep learning on low-dose CT. Nat Med. 2019;25(6):954–961.

[8] McKinney SM, Sieniek M, Godbole V, Godwin J, Antropova N, Ashrafian H, et al. International evaluation of an AI system for breast cancer screening. Nature. 2020;577(7788):89–94.

[9] Wang P, Berzin TM, Glissen Brown JR, Bharadwaj S, Becq A, Zhao X, et al. Real-time automatic detection system increases colonoscopic polyp and adenoma detection rates. Gut. 2019;68(10):1813–1819.

[10] Nemati S, Holder A, Razmi F, Stanley MD, Clifford GD, Buchman TG. An interpretable machine learning model for accurate prediction of sepsis in the ICU. Crit Care Med. 2018;46(4):547–553.

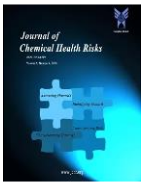
[11] Ting DSW, Cheung CY, Lim G, Tan GSW, Quang ND, Gan A, et al. Development and validation of a deep learning system for diabetic retinopathy and related eye diseases using retinal images from multiethnic populations with diabetes. JAMA. 2017;318(22):2211–2223.

[12] Shu T, Zhang B, Tang YD. A 10-year risk prediction model for type 2 diabetes based on routine clinical laboratory tests. Front Endocrinol. 2022;13:1060718.

[13] Hannun AY, Rajpurkar P, Haghpanahi M, Tison GH, Bourn C, Turakhia MP, et al. Cardiologist-level arrhythmia detection and classification in ambulatory electrocardiograms using a deep neural network. Nat Med. 2019;25(1):65–69.

[14] Topol EJ. Welcoming new medical imaging datasets. Nat Med. 2023;29(7):1581–1582.

[15] Rieke N, Hancox J, Li W, Milletari F, Roth HR, Albarqouni S, et al. The future of digital health



with federated learning. NPJ Digit Med. 2020;3(1):119.

[16] Li L, Qin L, Xu Z, Yin Y, Wang X, Kong B, et al. Artificial intelligence distinguishes COVID-19 from community acquired pneumonia on chest CT. Radiology. 2020;296(2):E65–E71.

[17] Moor M, Banerjee O, Abad ZSH, Krumholz HM, Leskovec J, Topol EJ, et al. Foundation models for generalist medical artificial intelligence. Nature. 2023;616(7956):259–265.