Artificial Intelligence Applications for Early Disease Detection in Healthcare Systems

Pooja Gautam[1*], Dr. Valli Madhavi Koti[2], Ms. Chanchal Garg[3], Dr. Reetika Agarwal[4], Dr. M. Saravanan[5], Mr. Shivendra Varshney[6]

[1*]Assistant Professor, Department of CSE, GL Bajaj Institute of Technology & Management, Greater Noida, Uttar Pradesh, India | Email: poojamkrsm@gmail.com

[2]Professor, Department of Computer Science & Engineering, Godavari Global University, Rajamahendravaram, Andhra Pradesh, India | Email: vallimadhavi@ggu.edu.in

[3]Assistant Professor, Department of CSE(DS), Noida Institute of Engineering and Technology, Greater Noida, Uttar Pradesh, India | Email: chnchlgarg4@gmail.com

[4]Associate Professor, Department of Management, IILM Academy of Higher Learning, Lucknow, Uttar Pradesh, India | Email: reetikaagarwal82@gmail.com

[5]Professor, Department of ECE, IFET College of Engineering, Villupuram, Tamil Nadu, India | Email: saravana_vkms@yahoo.com

[6]Assistant Professor, Department of Computer Engineering and Applications, Mangalayatan University Beswan, Aligarh, Uttar Pradesh, India | Email: shivendra.varshney@mangalayatan.edu.in

***Corresponding Author:** Pooja Gautam, Assistant Professor, Department of CSE, GL Bajaj Institute of Technology & Management, Greater Noida, Uttar Pradesh, India | Email: poojamkrsm@gmail.com

Abstract

With delayed access to specialist expertise and subjective interpretation by clinicians and workload limiting the ability to access parts of the pathway, early disease detection is one of the most impactful levers that can be used to improve patient outcomes and reduce the cost of care. Medical image analysis, electronic health records (EHR), genomic data analysis, and analysis of physiological signals are all examples of medical data that can be transformed by artificial intelligence (AI) and especially machine learning (ML) and deep learning (DL), as there is no way they could be analyzed at the same scale and speed as would be possible by a human being. In this paper we bring together existing evidence of the use of AI in early detection in oncology, cardiology, neurology, dermatology, ophthalmology and critical care. It discusses the mathematical approaches used for their analysis such as the use of convolutional neural networks for image analysis and recurrent networks for time-series analysis of physiological data, and compares their reported diagnostic performance with that of conventional clinical analyses. The paper also examines real-world challenges to adoption, such as standardization of data, algorithmic bias, regulatory approval processes and clinician acceptance, and proposes some solutions. The results suggest that AI systems perform as well as or better than humans in precisely defined detection tasks but will not automatically translate this to better population health outcomes without attention to data quality, the need to integrate AI systems into workflows, and the need to ensure that everyone has access to them. The paper ends with policy, research and system recommendations for maximizing early detection programmatic value while responsibly scaling these initiatives.

Keywords - Artificial Intelligence, Machine Learning, Deep Learning, Early Disease Detection, Medical Imaging, Electronic Health Records, Algorithmic Bias, Clinical Decision Support.

How to cite this article: Gautam P, Koti VM, Garg C, Agarwal R, Saravanan M, Varshney S. Artificial Intelligence Applications for Early Disease Detection in Healthcare Systems. Int J Drug Deliv Technol. 2026;16(70s): 1311-1317. DOI: 10.25258/ijddt.16.70s.140

1. Introduction

Traditionally, diagnosis of disease has depended on clinical skills, laboratory analyses and diagnostic imaging, all of which are performed and reviewed manually. This model is effective, but it is under increasing strain due to greater patient flow, a shortage of specialists (radiologists, pathologists and others) worldwide, and the fact that there are limits to the value of human judgment. The ability to make the correct diagnosis is a major cause of patient harm, and missed diagnoses of conditions like cancer, cardiovascular disease and neurodegenerative disorders often lead to poor outcomes and increased healthcare expenses.

There's a complementary way to it, using artificial intelligence. AI is able to extract patterns from vast amounts of labeled medical data, even before they are clinically visible or noticed by human eyes, and build statistical models that can identify patterns related to disease states. They are now in use in virtually all the main clinical disciplines, such as automatic grading of diabetic retinopathy in ophthalmology and sepsis-risk alerting in intensive care units.

This paper gives a comprehensive, structured review on the use of AI for early disease detection, evidence for clinical utility, and challenges to overcome for widespread, safe and equitable use. The goals of this review are threefold: to review the fundamental methodologies used in the field of AI for diagnosis; to review applications, categorized by their major disease area, and provide representative performance data; and to discuss the operational, ethical and regulatory challenges with the application of AI in a clinical setting for diagnosis.

2. Background and Significance

2.1 The Case for Early Detection

For most disease categories, earlier detection is strongly correlated with the survival rate, less invasive treatment and reduced overall cost of care. There are many types of cancer where the odds of surviving 5 years or more rapidly decreases with each advance in stage at diagnosis, with early diagnosis being one of the biggest factors in lowering cancer death rates in oncology. The same can happen with cardiovascular disease, where early detection of at-risk patient can provide a window to prevent their first heart attack; or with neurodegenerative diseases, which can be treated to reduce disease severity if the patient is detected early.

2.2 Conventional diagnostic pathways has its limitations.

The traditional diagnostic process relies on specialist skills and availability. With a high number of high volume imaging studies, radiologists can fall victim to fatigue-related error and may not have the ability to access subspecialist interpretation, especially in low resource and rural settings. There is also the throughput constraint of screening programs based on manual interpretation, as the number of people that can be screened realistically is limited.

2.3 AI's role in drug discovery and development.2.5 AI's contribution to personalizing healthcare.

In recent years, as computing power has increased, medical records have been digitised and large imaging datasets have been annotated to include more detailed labels, deep learning models have been able to perform on specific tasks in diagnostics, that they can match or outperform trained specialists. AI-powered software as a medical device (SaMD) has emerged as a rapidly expanding category with more products approved by regulatory authorities like the U.S. Food and Drug Administration as diagnostic aids.AI-powered software as a medical device (SaMD) is a growing category, and both the clinical and regulatory worlds are gaining confidence in these tools as diagnostic aids with more products gaining clearance from regulatory bodies like the U.S. Food and Drug Administration.

2.4 Scope and Approach of This Review

This paper adopts a narrative review methodology, summarizing in-depth, peer-reviewed research from the last few years, plus systematic reviews and industry reports surrounding AI-powered early disease detection. It's not just about theoretical research or work in the early stages of the lab... it's about applications that have been clinically validated or are emerging, and it takes a closer look at the cross-cutting themes of the research such as methodology, barriers to implementation and regulatory context, rather than trying to cover every published study. Specific performance numbers are quoted when they are provided, but these are not necessarily absolute, reproducible performance characteristics, but are rather representative of ranges quoted in the literature; the accuracy of the results reported varies considerably, depending on the composition of the data sets, study design, and clinical situation.

3. Principal Artificial Intelligence techniques applied in diagnostics applications.

3.1 Machine Learning and Deep Learning:

Machine learning is an umbrella term that refers to a family of statistical methods in which the patterns to be learnt from the data are not explicitly programmed into the machine. One of machine learning's newest subsets, deep learning, based on multi-layered artificial neural networks, represents the predominant method for processing unstructured data, such as images, textual information and physiological waveforms.

3.2 Convolutional Neural Networks for Medical Imaging

The architecture that is used the most for medical image analysis is called a convolutional neural network (CNN). Through the learning of hierarchical spatial features, CNNs can detect and recognize the subtle patterns in radiograph, CT scans, retina photos, and dermoscopic images, which relate to disease states. CNN based systems have been used in the fields of lung nodule detection in ct, tumour detection on mammography, and lesion classification for dermatology.

3.3 Sequence Models for Physiological and EHR Data

Typical examples of time-series data include continuous vital signs, lab trends, and electrocardiogram (ECG) waveforms, which these types of models are often used on. The models are especially applicable to identifying a slowly changing process, such as predicting sepsis, or to predict cardiovascular risks from longitudinal information from an EHR. Moreover, convolutional architectures like MobileNetV2 have also been optimized for use in low-power embedded and mobile devices so as to achieve real-time ECG classification on wearable devices in the home environment (PMC, 2022a). Previous research has focused on using machine learning directly in the triage process in the ED to identify patients who should be sent home without an ECG, based on the structured features found in the EHR that are relevant to this specific task and are available within a short time (PMC, 2022b).

3.4 Natural Language Processing

Natural language processing (NLP) technologies identify the underlying meaning of unstructured clinical content, such as physician notes, radiology reports and pathology reports. NLP is increasingly being applied to find undocumented risk factors, flag up any findings to follow up and to aid risk stratification for population groups.

3.5 Multimodal and Federated Approaches

The use of imaging, genomic, laboratory and clinical text data, in a single multimodal model, is increasingly being utilized to enhance diagnostic accuracy in emerging systems. Federated learning is being adopted where models are trained in multiple institutions, without centralizing patient data, while aiming to increase diversity of training data while maintaining privacy.

4. Applications Across Disease Areas

4.1 Oncology

The use of early detection technology via AI imaging analysis is one of the most advanced applications (PMC, 2025b). In lung cancer screening, deep learning models have been shown to be able to detect suspicious nodules in low-dose computed tomography (LDCT) scans with an ability to do so that, in some studies, has been higher than that of the unaided radiologist. For breast cancer screening, CNN-based systems are used as an independent, or as a concurrent reader, next to a radiologist, and have been shown to decrease the rate of false negative reads.

4.2 Cardiovascular Disease

Longitudinal EHRs and ECG waveforms can be used to train AI models that can detect patients with high risk of cardiac events years before a clinical diagnosis would be made (PMC, 2025d). Using deep learning on standard 12-lead and single-lead ECGs has demonstrated and, in some cases, outperformed the performance of experts both in detection of arrhythmias and in the detection of asymptomatic left ventricular dysfunction in primary care, as seen in the pragmatic trials EAGLE (PMC, 2025a). While the number of studies that assess the performance of AI in acute myocardial ischemia is limited, there have been dozens of peer-reviewed research reports of its superior diagnostic accuracy and efficiency for triage as compared to traditional interpretation (PMC, 2025e). These models combine structured risk factors with signal level ECG features that are not easily understood by human clinicians to provide risk stratification that can help facilitate earlier intervention and prevention.

4.3 Neurology

Multimodal deep learning models using MRI and cognitive assessment information, as well as various biomarkers, have demonstrated good performance in predicting early changes in the brain and the presence of Alzheimer's disease and related neurodegenerative disorders, sometimes several years prior to disease onset. This “window of opportunity” allows the possibility for intervening to reduce disease progression.

4.4 Ophthalmology

One of the earliest autonomous AI diagnostic uses that has been FDA cleared is automated grading of DR from retinal fundus images. These help to allow retinopathy screening to be done in primary care without the need for a local ophthalmologist in attendance, greatly improving the access to screening for diabetics.

4.5 Dermatology

CNN-based classifiers can differentiate malignant from benign skin lesions, aiding in triage decisions regarding patients who need immediate specialist referral, when images are obtained with a dermoscope and smartphone. Current research is being directed on maintaining performance over the entire range of skin tones on the various presentations of lesions.

4.6 Critical Care and Sepsis Detection

Machine learning models in inpatient and intensive care wards constantly monitor trends in vital signs, lab results, and nursing documentation, alerting to important indicators of clinical deterioration, such as sepsis, often much sooner than would a human.

4.7 Emerging Multi-Cancer and Liquid Biopsy Applications

There is a newer generation of AI in diagnosti which uses machine learning and liquid biop technology, detecting circulating tumour DN and other cancer markers in blood to test for multiple cancers in one sample. Multi-cancer early detection (MCED) tests use laboratory assays and classification algorithms that are designed to detect methylation or mutati patterns associated with cancer, whi minimizing the background signals from the body. There is mixed evidence on how well it works in practice. The Galleri MCED test correctly identified the predicted cancer signal origin in the majority of true cancers in tl PATHFINDER 2 study, and had a low fals positive rate (GRAIL, 2025). The next, first-o its-kind, NHS-Galleri randomized controlled trial (RCT) failed to achieve its primary objective of lowering the number of patients diagnosed with stage III/IV cancer, although it did improve cancer detection and slightly lower the number of patients diagnosed with stage IV or in an emergency (ASCO, 2026; PMC, 2025c). This combined finding highlights that the potential benefits of using AI in diagnostics in the lab and early development stages do not necessarily extend to the population-level clinical outcomes that are measured in randomized trials, an issue that wasraised in Section 6.

4.8 Genomics and Pharmacogenomic Risk Prediction

AI is also being used in genomic data to predict inherited risk of disease earlier – even before clinical or radiographic evidence – and to provide this information in a way that is easy to interpret.AI is also used to apply this to genomic data to predict inherited risk of disease prior to clinical or radiographic evidence and to present this information in an easy-to-understand format. The polygenetic risk scores are predictive scores that estimate a person's relative risk of developing a disease (e.g. coronary artery disease, type 2 diabetes, and some cancers) that are derived from machine learning models trained on large genomic cohorts that rely on the accumulated effect of a large number of genetic variants. In addition to clinical risk factors these scores can assist in determining patients who may need more intensive screening than would be considered with the standard practice guidelines.

Table 1. AI Techniques Applied to Early Detection by Disease Area

Disease	AI Technique	Data Type	Benefit
Lung Cancer	CNN	CT / X-ray	Higher nodule sensitivity
Diabetic Retinopathy	DL classifier	Fundus photos	Enables mass screening
Breast Cancer	CNN + ensemble	Mammography	Fewer false negatives
Cardiovascular	Boosted models, RNN	ECG, EHR	Early risk prediction
Skin Cancer	CNN (dermoscopic)	Dermoscopic images	Specialist-level triage
Alzheimer's	Multimodal DL	MRI, biomarkers	Detection pre-symptom
Sepsis / ICU	LSTM, XGBoost	Vitals, EHR streams	Earlier alerting

Table 1 summarizes representative AI methodologies, data modalities, and reported clinical benefits across major disease categories discussed in Section 4.

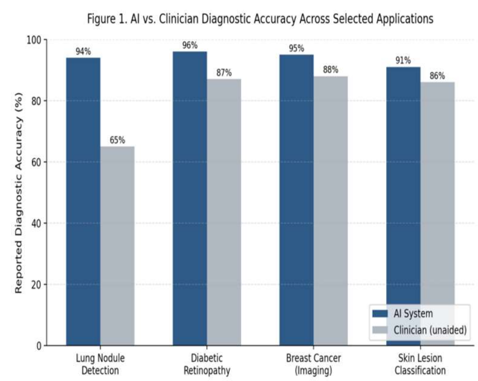

Figure 1 compares reported diagnostic accuracy of AI systems against unaided clinician performance for four representative applications. Figures are illustrative of ranges reported in the literature and vary by study design, dataset, and clinical setting.

5. Adoption Rates and Market Share for Different Adoption Levels

In the past few years, there has been a significant increase in investment in AI-powered diagnostics, with evidence of clinical use, increasing regulatory clearance routes, and interest from health systems to enhance diagnostic accuracy and efficiency of their staff. AI-powered healthcare diagnostics are expected to continue growing at a rapid pace throughout the decade, as both the number of vendors and procurement by hospital systems and payers grows.

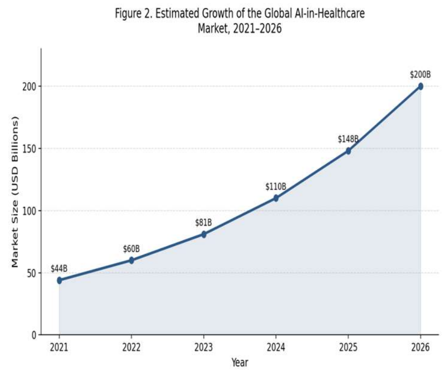

Figure 2 illustrates the estimated growth trajectory of the global AI-in-healthcare market (all clinical and administrative applications, not diagnostics alone), backed out from industry reporting that the market exceeds approximately USD 200 billion by 2026 at an annual growth rate above 35%. Values are directional estimates aggregated from industry market research rather than a single authoritative source.

Specialties that rely heavily on imaging, like radiology and ophthalmology, have seen the highest adoption rates, because the pathways to and outcomes of imaging are well established in these areas. The adoption of solutions where multiple modalities are needed, such as sepsis prediction and neurodegenerative disease risk scoring, has been slower, as the validation of models in heterogeneous clinical settings has been more complex.

6. Challenges and Limitations

Although these AI diagnostics have shown promising results in clinical tests, there are unique operational, technical, and ethical complexities when implementing them in clinical practice.

6.1 Data Quality and Standardization

AI models need to be trained with lots of good-quality data, which is consistently labeled. In reality, there are significant differences among eHR formats, imaging protocols and coding standards across institutions, which makes it difficult to train models that will apply to a wider variety of settings than their development context.

6.2 Human Rights, Equity and Diversity in the Public Sector.

A training set may be biased towards a specific population (differences in race, sex, age, and/or socioeconomic status) and yield models with poorer performance for these groups. A systematic review of AI and racial disparities in healthcare revealed a consistent pattern of worsening racial disparities when AI is used, which was attributed to biased training data, algorithm design choices, and inequitable practices in implementing AI in healthcare systems (Journal of Racial and Ethnic Health Disparities, 2024). For instance, a commercial risk-prediction model was determined to under-predict the need for further chronic-disease care for Black patients compared to White patients with similar clinical risk, after adjusting for bias (Yale School of Medicine, 2024). To ensure that AI diagnostic tools are fair for all populations, it is crucial to pay attention to and actively monitor performance by subgroups.

6.3 Regulatory and Validation Pathways

The regulation of software as a medical device is ongoing, especially when software is adaptive and is able to adapt itself after deployment. However, multi-site prospective validation studies are still required to prove that the performance seen in retrospective studies is applicable to real world clinical settings.

6.4 Clinical Workflow Integration

It's important to remember that while AI may be accurate, if its integration into clinical workflows is not well designed, it can fail to have a positive impact on efficacy. Inadequately designed alerting systems have the potential to increase clinician alert fatigue and systems that require extra effort on the clinician's part to use are unlikely to be adopted.

6.5 Explainability and Clinician Trust

There are numerous deep learning models that perform very well that are essentially a black box and it is hard to determine what is the basis for a specific prediction. The lack of interpretability can lead to reduced clinicians' trust and make it more difficult to hold them accountable if the AI-assisted decisions are subsequently reviewed.

6.6 Data Privacy and Security

The information shared about health is very sensitive, and AI systems that are capable of processing a tremendous amount of patient data are an easy target for data breaches. Strong encryption, access restrictions and anonymization measures are vital aspects of any AI deployment.

Table 2. Key Implementation Challenges and Mitigation Strategies

Challenge	Issue	Mitigation
Data quality	Inconsistent formats across sites	Federated learning; common models
Algorithmic bias	Underrepresented groups in data	Diverse data; subgroup audits

Regulatory approval	Evolving SaMD frameworks	Early engagement; validation
Clinical integration	Alert fatigue, workflow disruption	Human-centered design
Explainability	Black-box model outputs	Interpretable ML; saliency maps
Data privacy	Breach and misuse risk	Encryption; access controls

Table 2 outlines common barriers to real-world AI deployment in diagnostic settings alongside strategies that have been proposed or adopted to address them.

7. Quality and the Code of Conduct for Professional Services

The use of AI for early detection of disease doesn't only bring up technical issues, but also questions about the nature of the research. The use of AI in the diagnostic process requires informed consent, which should take into consideration the role of AI in the diagnostic journey. Responsibility in the context of liability frameworks needs to be clear in cases of a diagnostic error being caused by an AI-assisted decision. Oversight bodies have to strike a balance between the need to make useful tools available in a timely fashion and to have on-going intense safety monitoring.

There is an increasing number of recommendations and guidance from regulatory bodies and professional organizations for ongoing post-market performance monitoring, reporting of the characteristics of training data, and methods by which clinicians and patients can be made aware of when and how AI has played a role in a diagnostic decision.

8. Future Directions

The future of early disease detection with AI shows promise of several trends. First, multimodal models, which combine imaging, genomic, laboratory, and clinical text data, are anticipated to perform better than single-modality models, as it is hoped to be a more comprehensive understanding of the risk of individual patients. Second, federated learning techniques are likely to grow in order to train models on larger, more varied sets of data without having to consolidate sensitive patient information in a single location. Third, explainable AI will help increase clinician trust and help with regulatory review. Lastly, the increasing use of AI in consumer-facing applications and devices, such as wearable technology and mobile screening programs, will take early detection beyond the clinic and into real-world settings.

To achieve this potential, investments in data infrastructure will need to be maintained, regulatory pathways must be further developed to accommodate adaptive AI systems, and technologists, clinicians, and policymakers must continue to work together to ensure these tools are used to support – not hinder – patient care.

9. Healthcare Stakeholders recommendations

To deliver the diagnostics potential of AI to clinical benefit in a reliable and equitable way, we must work together as a health system, researchers and policymakers. The evidence and challenges discussed above are summarised in the following recommendations, which are priorities for each group.

9.1 For Health Systems and Clinical Leaders

- Test AI diagnostics tools in clinical workflows on a smaller scale, prior to full rollout and assess actual performance in real-world settings as well as vendor-provided accuracy.
- Set up subgroups for performance monitoring to identify and address inaccuracies between patient subgroups.
- Provide clinician training to ensure AI-generated outputs are used as part of a clinician's decision making process, not as the clinician's own unquestioned decision.

9.2 For Researchers and Developers

- Avoid retrospective single institution studies that often overestimate real world results, in favor of prospective, multi-site validation studies.
- Document training data demographics and known limitations clearly and openly to enable informed clinical usage.
- Implement strategies to increase explainability for clinicians to gain insight into the drivers behind a model's output, especially when making important clinical decisions about a diagnosis.

9.3 For Policymakers and Regulators

- Continue to develop regulatory frameworks appropriate to adaptive, learning AI systems, instead of AI systems that are once-and-for-all approved devices.
- Enable data-sharing frameworks and infrastructure including federated learning networks that increase diversity of training data without compromising patient privacy.
- Pro-collaborate on reimbursement and coverage policies that incentivize better diagnostic results instead of just adoption of an AI tool in the workflow.

10. Conclusion

From cancer imaging to cardiovascular risk prediction to neurodegenerative disease identification, AI has proven to have significant promise in advancing the field of early disease detection in various clinical areas. The findings included here suggest that, at the level of a very specific diagnostic task, AI systems can perform at the same level or better than the human clinical review process. However, a number of implementation issues must be overcome to achieve the full population health benefit these tools provide, such as data standardization, algorithmic bias, regulatory validation, workflow integration and clinician trust. The healthcare organizations that take a proactive approach to these core investments are most likely to make the most impactful and equitable contribution to patient outcomes, leveraging the diagnostic potential of AI.

References

1. Alnaggar, O. A. M. F. (2024). Efficient artificial intelligence approaches for medical image processing in healthcare: Comprehensive review, taxonomy, and analysis. Artificial Intelligence Review, 57(8).
2. Alshraideh, M. (2024). Enhancing heart attack prediction with machine learning: A study at Jordan University Hospital. Applied Computational Intelligence and Soft Computing, 2024.
3. American Society of Clinical Oncology (ASCO). (2026). Galleri early detection test may shift timing of cancer detection, reducing stage IV diagnoses.
4. GRAIL, Inc. (2025). PATHFINDER 2 results show Galleri multi-cancer early detection blood test increased cancer detection more than seven-fold when added to USPSTF A and B recommended screenings.
5. Journal of Chemical Health Risks. (2026). Role of artificial intelligence in early disease diagnosis: A narrative review. Vol. 16, No. 2.
6. Journal of Racial and Ethnic Health Disparities. (2024). The algorithmic divide: A systematic review on AI-driven racial disparities in healthcare.
7. Journal of World Future Medicine, Health and Nursing. (2026). Artificial intelligence in early disease detection: Revolutionizing diagnostic practices in medicine. Vol. 4, No. 1.
8. MDPI Applied Sciences. (2026). Artificial intelligence in medical diagnostics: Foundations, clinical applications, and future directions. Appl. Sci., 16(2), 728.
9. National Institutes of Health. (2026). AI systems and diagnostic sensitivity in lung cancer and diabetic retinopathy screening.
10. PMC (National Center for Biotechnology Information). (2022a). Artificial intelligence for cardiac diseases diagnosis and prediction using ECG images on embedded systems.
11. PMC (National Center for Biotechnology Information). (2022b). Development and validation of an artificial intelligence electrocardiogram recommendation system in the emergency department.
12. PMC (National Center for Biotechnology Information). (2025a). Artificial intelligence in electrocardiography: From automated arrhythmia detection to predicting hidden cardiovascular disease.
13. PMC (National Center for Biotechnology Information). (2025b). Artificial intelligence in radiology: Advancing precision, accuracy, and early detection in cancer diagnosis.
14. PMC (National Center for Biotechnology Information). (2025c). Clinical referral to the NHS following multi-cancer early detection test results from the NHS-Galleri trial.
15. PMC (National Center for Biotechnology Information). (2025d). Systematic review of artificial intelligence and electrocardiography for cardiovascular disease diagnosis.
16. PMC (National Center for Biotechnology Information). (2025e). The role of artificial intelligence in improving diagnosis, management, and outcomes of acute myocardial ischemia: A systematic review.
17. U.S. Food and Drug Administration. Artificial intelligence and machine learning (AI/ML)-enabled medical devices.
18. World Health Organization. Ethics and governance of artificial intelligence for health: Guidance.
19. Yale School of Medicine. (2024). Eliminating racial bias in health care AI: Expert panel offers guidelines.