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The Role of Artificial Intelligence in Diagnostic Medicine: A Narrative Review

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Abstract

Artificial intelligence (AI) has been with us since the 1950s and has long since undergone major developments in its capabilities and complexities, but it has recently evolved to a point where its capabilities have been substantially enhanced, all thanks to technological advances in the field of AI and modern hardware. While AI has already found applications in medicine, new revolutionary uses for it are emerging that could profoundly impact the future of healthcare for both clinicians and patients alike. However, with every new technology comes with it new issues to tackle, which can pose significant challenges. Thus, this paper aims to explore the following: what is AI and how does it function (a), its role in medical imaging (b), its application in predictive diagnostics (c), its future in genomics and personalized medicine (d), its contributions to personalized medicine (e), its limitations that require addressing (f) and its future in the field of medicine (g).

Subject Areas

Healthcare & Technology

Keywords

Artificial Intelligence, Machine Learning, Diagnostic Medicine, Technology, Narrative Review

1. Introduction

The rapid evolution of artificial intelligence (AI) over the past several decades has transformed many industries, including healthcare. From its early beginnings in the 1950s, AI has developed into a sophisticated technology capable of complex tasks, making it an increasingly valuable asset to the medical sector. With its ability to process and analyze vast datasets, identify patterns, and offer predictive insights, AI is setting new standards within the sphere of medicine. As modern healthcare shifts towards precision medicine and personalized healthcare, AI is well-positioned to revolutionize healthcare systems by enhancing the accuracy and improving the efficiency of clinicians. However, the integration of AI into healthcare also presents several challenges, including ethical and legal concerns along with the potential biases of AI systems. As AI technology further advances and integrates into medicine, it is this paper's objective to explore the multifaceted role of AI in medical imaging, its potential in predictive diagnostics and genomics and its future in personalized medicine along with the limitations of integrating AI into the field of medicine.

2. Methods

The information in this narrative review has primarily been derived from research articles and reviews published in scientific journals within the past 5 years (2019-current). Some information has been referred from websites pertaining to organizations that are affiliated with the development of AI technologies. Search engines and databases such as PubMed, ClinicalTrials and GoogleScholar were used to search for publications in the English language relevant to the topic of this paper. Search inputs included "AI in healthcare", "AI in diagnostics", "Machine-learning in medicine", etc.

3. Overview of Artificial Intelligence

Artificial intelligence refers to a system or algorithm within a computer that is capable of mimicking human rationale by learning and applying this algorithmic-based form of rationalization to complex processes such as problem-solving, decision-making, creativity and autonomy [1]. The very first program that utilized a form of artificial intelligence came about in 1952, and it was capable of partaking in games of checkers [2]. Since then, artificial intelligence has come a long way in its development and capabilities. Today, there is almost no segment of society that is completely devoid of AI or its effects—from the internet feeds and social media applications we are exposed to the automated machinery that produces our food to the security surveillance systems that are enforced within states—every imaginable aspect of modern society has been in some way affected by AI [3].

3.1. How Is AI Trained?

Artificial intelligence utilizes an array of different forms of technology that have been and continue to be in development till today. Two primary concepts of AI training include "machine learning" and "deep learning". Newer emerging technologies, such as generative AI, may utilize a blend of these two learning concepts [1].

3.2. Machine Learning

Machine learning (ML) involves inputting data into "models" with the goal of the models learning from these databases. This technique is very commonly utilized in training AI as it is the core to familiarizing AI with swathes of information [4]. There are three primary forms of ML:

1) Supervised Learning: Algorithms are trained on labeled data or data with predictable outcomes. For example, training model A to detect and treat patients with disease B [5].

2) Unsupervised Learning: Algorithms are trained to analyze and detect patterns of data within clusters of unlabeled datasets. For example, identifying novel risk factors within groups of patients that share a particular disease [6].

3) Reinforced Learning: Algorithms are trained to respond to the environment within the context of achieving a specific objective. For example, robotics in surgery where the AI experiences "positive feedback" when correct action is taken [7].

3.3. Deep Learning

Deep learning (DL) is a specialized subset of ML that involves having algorithms process through multiple layers of data where concepts are established on top of previously managed concepts (hence "deep"). This technique tries to replicate the way the human mind learns by utilizing neural networks with respective datasets. Algorithms trained on this technique excel in processing vast amounts of complex and high-dimensional data, making it an appropriate algorithm training technique within the context of medicine [4]. The first algorithm to utilize DL was in 1965, therefore, it is not a new technique. However, it has enjoyed tremendous developments within the past few years as a result of advancements made in computing and hardware [8].

4. AI in Medical Imaging

4.1. Radiology

Artificial Intelligence is an integral tool for radiologists in nearly every subspecialty of radiology. It plays a pivotal role in the detection and characterization of diseases seen in radiological images and has become integrated into many modalities of imaging. Aside from its diagnostic benefits, it is also capable of enhancing radiological images, thereby reducing scan times of MRI and CT, ultimately reducing exposure to radiation for both patients and operators alike [9].

Diagnostically, AI has been utilized in various unique subspecialties, ranging from neuroradiology to musculoskeletal radiology. Through DL, AI algorithms can be enabled to categorize and classify various abnormal elements that may be presented in a radiological image, such as hemorrhages, lesions, air, coronary plaques and fractures. As a result, it is also capable of identifying certain anatomical structures that act as waypoints for radiologists when they are assessing images, thereby increasing their efficiency and potentially the accuracy of their diagnosis [10].

Table 1 provides a brief overview of the multiple applications of AI in different subspecialties of radiology.

Table 1. An overview of the different diagnostic applications of AI in different subspecialties of radiology [9].

Field of activity/ subspecialty	Applications
Abdominal radiology	Segmentation of the liver, genitourinary structures, and other anatomical regions; identification of lesions, incidental detection of pulmonary emboli in the lung bases, free intraperitoneal air, vertebral compression fractures, or aortic dissection, characterization of focal liver and adrenal lesions.
Neuroradiology	Detection of lesions, identification of strokes and hemorrhages, detection of aneurysms, evaluation of brain anatomy, and segmentation and division of cortical and subcortical structures.
Chest radiology	Identification of lung nodules, detection of pneumonia, pneumothorax, and rib fractures, recognition of lines and tubes, diagnosis of obstructive lung disease, and quantification of emphysema.
Cardiovascular radiology	Coronary calcium scoring, coronary angiography, fractional flow reserve measurement, plaque analysis, left ventricular myocardium evaluation, diagnosis of myocardial infarction, prognosis of coronary artery disease, assessment of cardiac function, and diagnosis and prognosis of cardiomyopathy.
Musculoskeletal radiology	Identification of fractures in the proximal humerus, hand, wrist, and ankle, detection of hip osteoarthritis, and quantitative bone imaging for evaluating bone strength and quality.
Oncologic imaging	Tumor characterization and segmentation, identification of pulmonary nodules, tumor delineation, staging and classification of lung cancer, and distinguishing between benign and malignant lesions or lymph nodes.
Breast radiology	Detection, classification, and characterization of lesions, breast density estimation, characterization of mammographic abnormalities, and differentiation between malignant breast lesions and nodules.
Emergency radiology	Identification of intracranial hemorrhages, large vessel occlusions, fractures, free abdominal fluid, small bowel obstruction, and intussusception.
Urogenital radiology	Autonomic segmentation, whole-gland detection and zonal segmentation, lesion segmentation, volume estimation, tumor detection, cancer localization, and tumor grade assessment.
Head and neck radiology	Target delineation, segmentation of lesions and anatomical structures, lesion localization, lesion classification, and segmentation and classification of lymph nodes.
Oral and maxillofacial radiology	Target delineation, segmentation of lesions and anatomical structures, localization of lesions, classification of lesions, and segmentation and classification of lymph nodes.

4.2. Pathology

Within the context of pathology, AI is becoming an increasingly relevant tool to the interpretation of histological samples though it remains to be fully employed as a definitive diagnostic instrument [11]. With the advent of complex DL-based models, referred to as convolutional neural networks (CNNs), AI has become capable of recognizing the different structures within cells and recognizing certain patterns indicative of pathology. Through object recognition, detection and segmentation of elements within histological samples, AI can detect tumor cells and has shown the potential to assist in standardizing the scoring criteria for different types of tumors such as the Gleason score for prostate cancer—through representing the morphological changes seen within the different stages of a malignancy on a spectrum. Content-based image retrieval (CBIR), a system that is based on AI, can provide defined histological images as references from a repository that would match or appear similar to the histological samples that are inputted. This can drastically aid pathologists in their diagnoses, especially within the context of diagnosing complex or rare diseases [12].

4.3. Ophthalmology

Imaging in ophthalmology with the use of AI systems yields promising results though it is not a new technique. ML-based AI algorithms have been experimented with since as far back as 1976 in the CASNET-based glaucoma consultation program [13]. As ophthalmology is heavily image-based, AI has the potential to prove useful through image recognition. It has already shown potential in detecting diabetic retinopathy, glaucoma and age-related macular degeneration (ARMD). AI systems, particularly DL, are being further refined to analyze retinal images and detect subtle changes that often occur in the early phases of the aforementioned pathologies [14].

Diabetic retinopathy (DR), a leading cause of blindness worldwide, is a key example of where AI shows promise within the field of ophthalmology. The FDA approved the first AI-based algorithm (IDx-DR) for screening DR in 2018. This decision was based on its speedy and accurate screening capabilities, which non-ophthalmologists can take advantage of [15]. Other promising systems are currently being developed and employed which utilize a similar AI technology as to that of IDx-DR's AI algorithm. Such systems include Google's DeepMind and EyeArt [16].

In age-related macular degeneration (AMD), AI has proven useful in detecting early stages of the disease through the analysis of retinal images. By identifying characteristic signs such as drusen (yellow deposits under the retina), AI algorithms can help predict the progression of AMD and guide timely treatment interventions, thereby enabling a more accurate and efficient working strategy for clinicians [17].

Glaucoma has been detected in the past using AI models that are trained on optical coherence tomography (OCT) scans. These scans can detect structural changes in the optic nerve and retinal nerve fiber layer, both of which are key indicators of glaucoma progression [18].

5. AI in Predictive Diagnostics

Predictive analytics refers to the use of AI algorithms to analyze patient data with the purpose of forecasting future health outcomes. As AI models are particularly effective in identifying patterns that might not be immediately apparent to human clinicians, they have a massive potential to play in the field of predictive analytics. Predictive analytics would entail diagnosis, prognosis, risk assessment, treatment response, disease progression, readmission risks, complication risks and mortality prediction.

According to a 2024 study in the Journal of "Computer Methods and Programs in Biomedicine Update" on the role of AI in clinical prediction, the vast majority of studies on this topic were focused within the fields of oncology, radiology and neurology where AI yielded to be most useful in clinical prediction to oncologists and radiologists [19]. This result is understandable as both oncologists and radiologists alike, heavily rely on image scans in the diagnosis and treatment of their patients and AI has already proven to be effective in the interpreting of diagnostic radiological images.

An example of a notable application of AI in predictive analytics is in the detection of sepsis, a life-threatening condition characterized by the body's extreme response to infection. Sepsis is one of the leading causes of death in hospital intensive care units. ML-based AI models have been developed to predict the onset of sepsis by continuously analyzing patient data, such as heart rate, blood pressure, respiratory rate, temperature, and lab results. Though the AI technology behind this remains to be imperfect and in need of continued development before it can be fully deployed inside hospitals as a reliable instrument, it has already shown positive results in some studies [20]. An example of this success was seen in the "targeted real-time early warning system" (TREWS) developed by researchers at the John Hopkins University. The TREWS system is designed to detect sepsis in patients before the usual clinical symptoms become apparent, thereby notifying clinicians to promptly act and ultimately improve patient survivability before their condition worsens. In their study, TREWS was capable of detecting sepsis in up to 82% of patients before symptoms worsened [21].

AI is also widely used to predict complications in chronic diseases, such as heart failure, diabetes, and chronic obstructive pulmonary disease (COPD). Predictive models analyze data from electronic health records, lab results and patient-reported symptoms to forecast disease progression and identify patients at risk of hospitalization. For example, a study showed that an AI model trained on different datasets of electronic health records and comorbidities of COPD patients can predict the likelihood of 30-day hospital readmission in up to 87% of cases [22]. This type of information can help clinicians tailor specific treatment plans for their patients and enables a form of risk stratification.

Predictive AI in the treatment of cancers using immunotherapy has also shown promise. The "logistic regression-based immunotherapy-response score" (LORIS) is an AI tool that predicts the efficacy of immunotherapy in patients with cancer based on their age, cancer type, treatment history and several other factors. According to the creators of LORIS, the ML-based AI algorithm was able to consistently and accurately predict patient short-term and long-term survivability in a study consisting of around 4,000 patients [23].

6. AI in Genomics

Genomic Data Analysis

Genomic data analysis involves examining the entirety of an individual's genetic information (genome) to identify mutations, variations, and patterns that may be linked to diseases or traits. AI is particularly useful in analyzing large-scale genomic data because of its ability to sift through massive amounts of data within a short period of time. ML and DL-based AI algorithms are highly efficient in identifying mutations associated with genetic disorders. For instance, AI systems can scan entire genomes to detect single nucleotide polymorphisms (SNPs), copy number variations (CNVs), and other mutations that may contribute to diseases like cystic fibrosis, Huntington's disease, and various cancers. As patient records become electronic and accessible through a computer, it becomes easier and more convenient to employ such AI technologies within the real-world setting [24].

AI has also shown the ability to detect mutations specific to different types of cancer, thereby aiding in the screening, diagnosis, classification and treatment of cancers. For instance, several ML-based AI algorithms can detect the BRCA1/2 mutation, relevant within the context of breast cancer and other types of cancer. These AI-based tools have even been shown to be more accurate than experts [25].

Many of these AI-based tools/classifiers are open-source, meaning they are available to the public along with the databases they are trained on. This aids in their development while simultaneously providing a sense of transparency to any potential future researchers and users who wish to take part in developing these emerging AI-based technologies as they remain to be in the experimental and research phase [26].

Figure 1 provides an overview of the current impact AI has left in the field of genomics relevant to cancer (oncogenomics).

Applications	How AI is applied	Impact
Cancer diagnosis and monitoring	Genomic data can be analyzed by machine learning models to find patterns linked to cancer. These models can help in cancer recurrence probability prediction, subtype categorization, and early diagnosis.	AI-assisted early diagnosis and monitoring lead to more individualized and efficient treatment plans, which enhance patient outcomes.
Identifying at-risk populations	Large-scale genetic databases can be analyzed by AI algorithms to determine which people are more susceptible to a given disease, such as inherited disorders or complicated disease susceptibility.	Public health initiatives can be strengthened by implementing screening programs, preventive measures, and targeted interventions for populations that are at risk.
Classifying genetic variations	Genetic variants can be categorized and interpreted by machine learning algorithms, which can differentiate between potentially hazardous and benign mutations. Understanding the genetic foundation of diseases requires knowledge of this.	Precise categorization of genetic variants facilitates the diagnosis of hereditary illnesses, directs therapeutic choices, and expands our comprehension of the genetic foundations of ailments.
Predicting ancestry of a patient	AI systems are able to predict an individual's ancestral ancestry by analyzing genetic markers. To do this, the genetic profile is compared to reference datasets made up of various demographic groups.	Because various genetic variants and susceptibilities might be associated with particular populations, ancestry prediction holds potential implications in personalized medicine. It also helps with customized healthcare planning.

Figure 1. An overview of the different applications of AI in the field of oncogenomics [24].

7. AI in Personalized Medicine

Personalized medicine, also known as precision medicine, involves tailoring a specific intervention for each patient based on their unique medical portfolio. It takes into account their medical history, lifestyle, genetics, biomarkers, risk factors, laboratory results and many other notable factors related to their health, with the goal of supervising an appropriate intervention that puts their "medical uniqueness" into consideration. With the increasing rate of utilizing electronic health records and the increased reliance on technology within the medical field, there exists a feasibility to utilize AI by running patients' data through AI-based algorithms with the aim of aiding clinicians in providing targeted therapy plans and risk predictions to their patients [27].

Organizations such as HealthJoy, Google, Paige.AI and Tempus have heavily invested in AI technologies and have already released platforms/tools that are AI-driven and function on the basis of categorizing personal data to create personal interventions. HealthJoy, for example, released a mobile application called "Virtual Primary Care" that is powered by an AI algorithm that aids patients in connecting to specific healthcare services depending on the specific information they choose to share. Tempus, on the other hand, has released online services for healthcare providers that are AI-driven and focused on personalized medicine. Such services included hubs to input and categorize patient data into (which both clinicians and researchers can take advantage of), an automated reporting of medical images, genomic assaying and cancer testing. These emerging AI tools may act as significant catalysts in driving the advancement of personalized medicine, enhancing its precision and effectiveness [28].

One noteworthy aspect of personalized medicine that AI may revolutionize is pharmacogenomics. Pharmacogenomics is the study of how one's genome influences the effects of drugs and is therefore relevant within the context of personalized medicine. By pulling large sets of genomic data and pointing out trends and patterns, AI can apply its ML-based algorithms to predict pharmaceutical properties and ultimately enhance the process of drug discovery. For example, AI can enable the virtual screening of compounds in silico drug repositioning and identify genes for specific mutations that may lead to disease [29].

8. AI's Natural Language Processing in Healthcare

Natural language processing (NLP) is a ML-based technique that enables AI to understand and communicate spoken language by processing text or speech [30]. In healthcare, this would translate to facilitating interactions between patients and clinicians, analyzing unstructured data from electronic health records, predicting disease onset and progression and identifying patients within clinical trials and studies [31].

One of the most significant applications of NLP in healthcare is summarizing, sorting and categorizing patient information from electronic health records. NLP tools, such as Tempus, can process through large chunks of inputted data to filter out specific information when sought after. This provides healthcare workers with a comprehensive view of their patients' medical records in a concise and neatly presented format which would ultimately optimize their workflow [32].

From a patient's perspective, NLP tools, in the form of mobile applications, may prove to be more cost-effective, time-saving and less troublesome than in-person visits to clinics for referrals. Rather than booking an appointment for an initial referral and seeing through it, which could possibly take days, patients could instead share their medical information with a responsive AI entity, which will redirect them to the relevant and specific healthcare providers that would treat them. Through this method, patients would avoid the hassle of visiting multiple clinicians for referrals while ultimately being sent to the human clinician/s that will treat them without the extra added steps in between that may discourage a patient from seeking healthcare services in the first place. The patient information shared could be linked to a database which would simultaneously sort, categorize and summarize their information while remaining accessible to healthcare workers. HealthJoy's services are an example of this—albeit implemented on a much smaller scale [33].

From a healthcare provider's perspective, NLP-trained AI algorithms can provide them with pertinent information about their patients within a short period of time, which would help guide their medical consultations and allow them to hone on their patients' medical concerns. Aside from NLP's administrative role and enhancing qualities, NLP also plays a role in disease identification, diagnosis, and drug discovery/development, as all of the ML-based AI algorithms discussed previously share an NLP element in order to relay the information back to the user in a clear and comprehensive language rather than in code which would require a computer engineer to decipher [34]. This is a very vital observation to note as it translates to AI tools being feasible and user-friendly for clinicians.

9. The Challenges of Integrating AI into Medicine

The future direction of integrating artificial intelligence into healthcare is currently being held back by certain challenges and considerations that must first be addressed.

As the broad use of AI in healthcare would entail the utilization of several online databases for sensitive patient information, safeguards must be designed and enforced to deter any potential cyberhackers that would leak personal information. This endeavor would be far from an easy one to overcome as technology evolves, the apparatus that hackers rely on will too evolve, resulting in a tick-for-tack battle between hackers and security systems, therefore necessitating constant security updates and upkeep. Any shortcomings in this sphere could lead to legal scandals and public outcries that could discourage future attempts at AI integration into medicine [35] [36].

The bias of AI algorithms based on the diversity of the data they are trained on must also be minimized sufficiently before they can be fully deployed within the clinical setting. Due to the wide-encompassing nature of the field of medicine, there will be different AI algorithms suited to different tasks. As technology evolves, and further efforts are placed into the development of individual AI algorithms, each separate modality must meet the standards of the local health authorities before being accepted for wide use in hospitals and clinics. This matter could span from years to decades before being resolved, depending on the pace of development, research and legalistic proceedings [36].

Ethical and legal considerations are by far one of the most restricting factors in integrating AI into any field but more so in medicine where sensitive information and lives' hang in the balance. Transparency in the training methods of AI algorithms, the databases that they were fed, and their intricacies and shortcomings must be made apparent to the public, especially those AI models directly involved with patients. Legal accountability is, for example, a crucial matter for debate that must be addressed and answered. As this technology emerges, new regulatory and legal screening methods must be enacted that would satisfy the public, the providers (of these AI technologies) and those within the health sector before it can officially be adopted [36] [37].

Seamless integration of AI-based tools into the medical field for both clinicians and patients is also of vital importance. These tools must adapt to the current technology that is in-use and must not pose as monumental challenges in their implementation. They should remain user-friendly and widely compatible with many platforms/devices [38].

Figure 2 provides an overview of a few hurdles that we must overcome before completely integrating AI into healthcare.

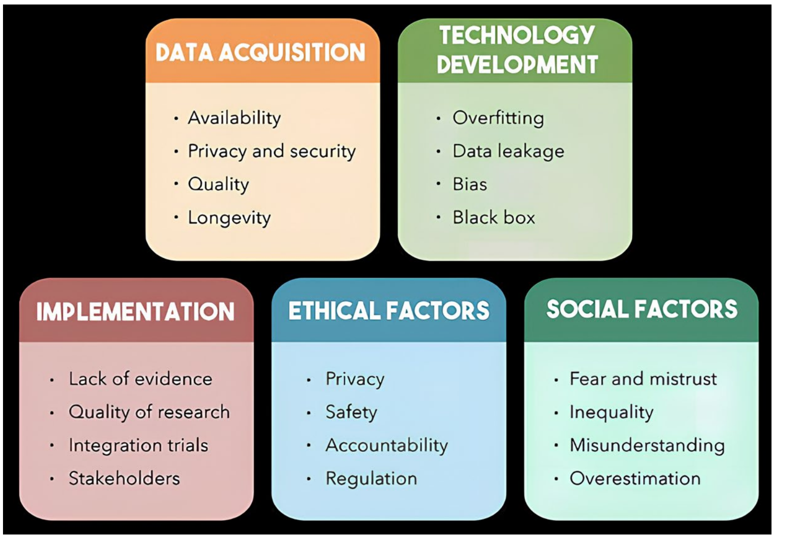

Figure 2. An overview of the issues of implementing AI that will require addressing [37].

10. Future Direction of AI in Medicine

It is difficult to assess the future direction of AI in every field of medicine. However, it is reasonable to suggest that its use for more administrative purposes, particularly through its natural language processing capabilities, will gain widespread adoption before its extensive application for diagnostic, monitoring and treatment purposes. This assumption is based on two factors. Firstly, many text-to-text NLP AI algorithms are already widely available for free or at low prices, making them easy to access and use for doctors and patients alike. Secondly, integrating AI systems into hospital devices for diagnosis, treatment or monitoring purposes will require time for development and legal approval, and financial investment for implementation. Though few of these technologies are already in use, particularly within the fields of radiology and surgery, AI remains by and large yet to be "fully" integrated into many specialties on a global scale.

According to a 2023 study conducted by the American Medical Association on the attitude of physicians towards AI, there appears to be an overall positive outlook on the role of AI in healthcare, with 65% believing it to be advantageous to them, while only 11% believing it to be disadvantageous. Out of the 1081 physicians surveyed, 38% were already using AI in their practice, not only for routine tasks like documentation and translation services but also for diagnostic purposes. 72% agreed that AI would most likely be helpful in its diagnostic abilities, while 41% expressed concerns about patient privacy. Three quarters of the respondents reported AI's documentation uses would be "most relevant to them" [39]. These statistics complement the notion of AI first being utilized for managerial purposes by physicians before being fully adopted as a modality to diagnose, treat, monitor or assess patients.

It is also worthy to note that there is currently no evidence suggesting AI will or possesses the potential to replace clinical specialists—rather, all evidence points towards it being/becoming a complementary tool to them [40].

11. Conclusion

In conclusion, significant advancements have been made in the development of AI technologies tailored to medical purposes. AI has demonstrated remarkable potential across various medical domains, including radiology, pathology, genomics, and personalized medicine. Its capacity to process vast amounts of complex data, identify patterns, and enhance diagnostic accuracy positions AI as a promising tool worthy of investment in time and research. However, the road to fully realizing AI's potential is fraught with challenges, including data privacy concerns, biases in algorithm training, and ethical considerations regarding transparency and legal accountability. Addressing these obstacles will be crucial for the safe and effective implementation of AI in healthcare.

Conflicts of Interest

The author declares no conflicts of interest.

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