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REVIEW ARTICLE

The Use of AI in Diagnosing Diseases: Present and Future

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ABSTRACT

This paper presents a comprehensive overview of the current and future applications of AI in disease diagnosis. Leveraging AI in healthcare has the potential to significantly enhance diagnostic accuracy, speed, and efficiency. The article highlights how AI-driven algorithms analyze medical images, such as X-rays, CT scans, MRIs, and mammograms, to expedite disease detection and diagnosis. Through deep learning models, AI showcases remarkable outcomes in identifying anomalies like tumors, fractures, and lesions, often surpassing human expertise. By furnishing preliminary findings, flagging areas of concern, and prioritizing urgent cases, AI assists radiologists in delivering expedited and precise diagnoses. Within the realm of clinical decision support systems, AI takes on a pivotal role. By analyzing expansive patient data encompassing genetic information, lifestyle factors, and medical history, AI can discern intricate patterns and risk factors linked to specific diseases. This early detection capability enables timely interventions and tailored preventive measures. AI-based risk assessment tools furnish personalized risk scores, further refining healthcare interventions. Additionally, AI-driven devices and wearable sensors perpetually monitor patients' vital signs, symptoms, and daily activities. These devices feed real-time data to AI algorithms, which promptly identify deviations, notify healthcare providers, and facilitate remote consultations. Particularly valuable for managing chronic diseases and expanding healthcare access to remote and underserved regions, this technology underscores AI's transformative potential in disease diagnosis and management.

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Introduction

The use of artificial intelligence (AI) in diagnosing diseases has been a significant area of research and development in recent years (Fig 1). AI has the potential to revolutionize the field of healthcare by improving diagnostic accuracy, speed, and efficiency (Davenport, 2019). Here's an overview of the present and future applications of AI in disease diagnosis

AI algorithms can analyze medical images such as X-rays, CT scans, MRIs, and mammograms to assist in the detection and diagnosis of various diseases (Fig 2) (Koh et al., 2022; Ghaffar et al., 2023). Deep learning models have shown promising results in detecting abnormalities, such as tumors, fractures, and lesions, often achieving accuracy levels comparable to or even surpassing human experts (Zhou et al., 2022; Meena & Roy, 2022). AI can help radiologists by providing preliminary findings, flagging areas of concern, and prioritizing urgent cases, leading to faster and more accurate diagnoses (Lee et al., 2019).

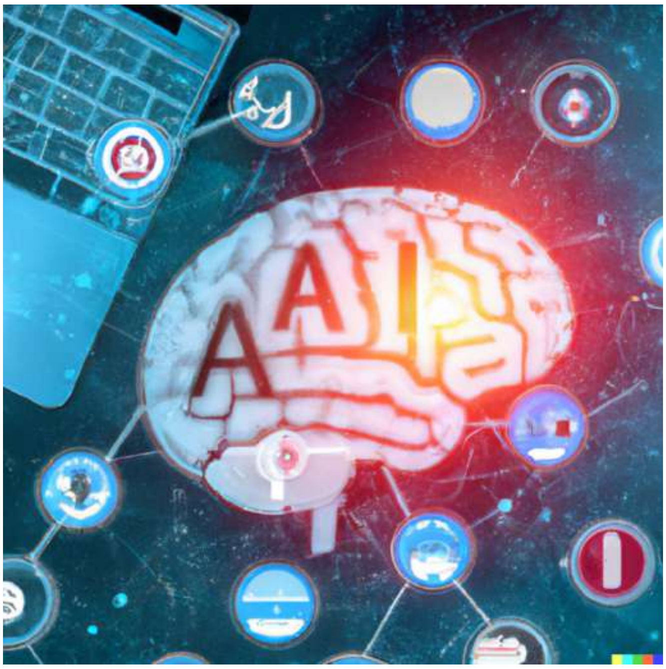

Fig 1. Development of AI.

AI algorithms can analyze digital pathology images, including tissue samples and slides, to identify patterns and markers associated with specific diseases, such as cancer (Baxi et al., 2022). This technology, known as computational pathology, can aid pathologists in detecting and classifying diseases, improving accuracy and reducing diagnostic errors (Bera et al., 2019). AI can also assist in quantifying biomarkers, predicting disease progression, and guiding personalized treatment decisions (Kumar et al., 2023).

AI has also made significant strides in the field of pathology and histopathology, offering new opportunities to improve disease diagnosis and treatment. Here's how AI is being utilized in pathology and histopathology (Kim et al., 2022).

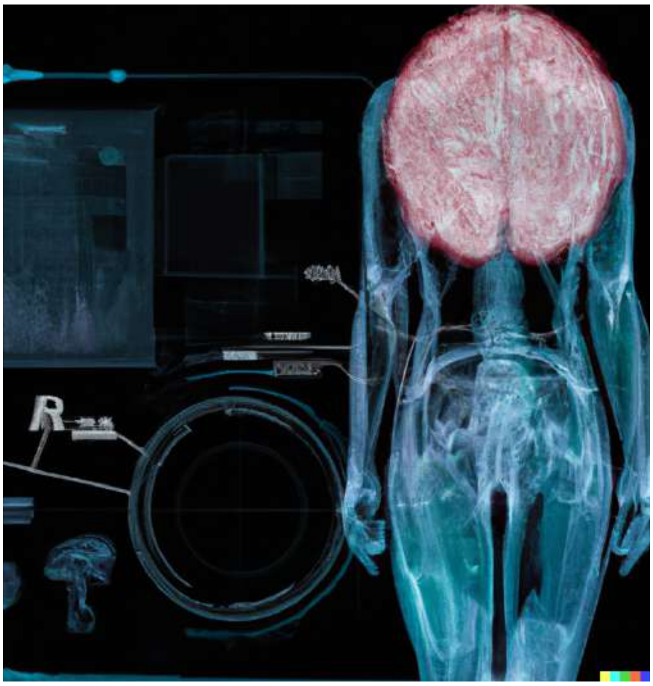

Fig 2. AI algorithms for X-rays, CT scans, MRI

AI can be integrated into electronic health records (EHRs) to provide clinical decision support systems (CDSS) (Wasylewicz & Scheepers-Hoeks, 2018). These systems can analyze patient data, including medical records, laboratory results, and genetic information, to suggest potential diagnoses, recommend appropriate tests, and provide treatment options (Sutton et al., 2020). CDSS powered by AI can help healthcare providers make more informed decisions, improve patient outcomes, and reduce medical errors (Sloane & Silva, 2020).

Clinical decision support systems (CDSS) powered by AI are computer-based tools that assist healthcare providers in making informed decisions about patient care (Sloane & Silva, 2020). These systems utilize AI algorithms to analyze patient data, medical knowledge, and evidence-based guidelines, providing recommendations and alerts to improve clinical decision-making (Moazemi et al., 2023). Here's how AI is utilized in clinical decision support systems:

AI models can analyze large amounts of patient data, such as genetic information, lifestyle factors, and medical history, to identify patterns and risk factors associated with specific diseases (Ghaffar et al., 2023). This enables early disease detection, allowing for timely interventions and preventive measures (Dhopte & Bagde, 2023). AI-based risk assessment tools can provide personalized risk scores, enabling healthcare professionals to tailor screenings and interventions to individual patients (Davenport & Kalakota, 2019).

AI-powered devices and wearable sensors can continuously monitor patients' vital signs, symptoms, and daily activities, providing real-time data for disease management. AI algorithms can analyze this data to detect changes or anomalies, alert healthcare providers, and facilitate remote consultations. This technology is particularly valuable for managing chronic diseases and improving access to healthcare in remote or underserved areas (Bohr & Memarzadeh, 2020; Yihan et al., 2023).

Applications

Medical imaging analysis

Medical imaging analysis using AI has emerged as a prominent application of artificial intelligence in healthcare (Tang, 2019). AI algorithms can analyze various types of medical images, including X-rays (Yoon et al., 2019; Azad et al., 2022), CT scans (Roberts et al., 2021), MRIs, and mammograms (Lei et al., 2021; Zheng et al., 2023), to assist in the detection, diagnosis, and treatment of diseases (Kufel et al., 2022; Kocak, 2022). Here are some key aspects of medical imaging analysis using AI:

Image interpretation

AI algorithms can be trained to interpret medical images and identify abnormalities or specific features associated with diseases. Deep learning techniques, such as convolutional neural networks (CNNs), are commonly used for image analysis (Yamashita et al., 2018; Yadav & Jadhav, 2019; Sarvamangala & Kulkarni, 2022). These algorithms learn from vast amounts of labelled data to recognize patterns and make accurate predictions (Hosny et al., 2018; Puttagunta & Ravi, 2021).

Automated detection and segmentation

AI can automate the process of detecting and segmenting structures or lesions within medical images (Sharma & Aggarwal, 2010; Rana & Bhushan, 2023). For example, AI algorithms can identify tumors, nodules, or anatomical structures like blood vessels or organs (Bi et al., 2019; Fahmy et al., 2022). This helps in identifying regions of interest and assists radiologists in making diagnoses.

Improving accuracy and efficiency

AI-based image analysis can enhance diagnostic accuracy and efficiency (Van et al., 2022). By aiding radiologists in identifying potential abnormalities or providing preliminary findings, AI can reduce human errors and help prioritize urgent cases. It can also speed up the interpretation process, leading to faster diagnoses and reducing patient waiting times (Van et al., 2022).

Screening and early detection

AI can be utilized in population-wide screening programs to detect early signs of diseases. For instance, AI algorithms have shown promise in the early detection of breast cancer through the analysis of mammograms. This enables timely interventions and improved patient outcomes (Cè et al., 2022; Uzun Ozsahin et al., 2022).

Quantitative assessment

AI algorithms can provide quantitative assessments of medical images, aiding in disease staging, treatment planning, and monitoring of disease progression. By analyzing the size, shape, and characteristics of lesions, AI can provide objective measurements that assist clinicians in making informed decisions (Hosny et al., 2018; Kumar et al., 2023).

Integration with clinical workflows

AI-based image analysis systems can be integrated into existing clinical workflows, including picture archiving and communication systems (PACS) and electronic health records (EHRs). This enables seamless access to AI-powered tools and findings, facilitating collaboration between radiologists and other healthcare professionals (Lee et al., 2021; Blezek et al., 2021).

Data-driven research and innovation

The analysis of large-scale medical imaging datasets using AI can unlock valuable insights and support medical research. AI algorithms can help identify novel biomarkers, discover new patterns or correlations, and contribute to the development of predictive models for disease prognosis and treatment response (Ghaffar Nia et al., 2023; Ahuja et al., 2019).

While AI has shown remarkable potential in medical imaging analysis, it is important to validate and refine these algorithms with rigorous testing and clinical studies. Collaboration between AI experts, radiologists, and other healthcare professionals is crucial to ensure the successful integration and responsible use of AI in medical imaging, ultimately improving patient care and outcomes.

Pathology and histopathology

Automated analysis of digital pathology images

Pathologists often analyze tissue samples and slides under a microscope to diagnose diseases. AI algorithms can process digital pathology images, which are high-resolution scans of these slides, and assist pathologists in identifying and characterizing diseases. Deep learning models, such as CNNs, can learn from vast image datasets and detect patterns and features associated with specific diseases.

Disease detection and classification

AI can aid in the detection and classification of diseases in pathology slides. By analyzing cellular structures, tissue architecture, and biomarkers, AI algorithms can identify abnormalities, such as cancerous cells or precancerous lesions. This can help pathologists make more accurate and efficient diagnoses, reducing diagnostic errors and providing better patient care (Wong et al., 2022).

Quantitative analysis and biomarker quantification

AI algorithms can perform quantitative analysis on pathology images, measuring characteristics such as cell counts, sizes, and distributions. This enables objective assessments and can be valuable in disease staging, treatment planning, and monitoring of treatment response. AI can also aid in the quantification of biomarkers, such as protein expressions, which can provide insights into disease behavior and guide personalized treatment decisions (Bera et al., 2019).

Computer-assisted grading and prognosis

AI algorithms can assist in the grading and prognostication of diseases based on pathology slides. For example, in cancer pathology, AI can help determine the grade of tumors, assess tumor aggressiveness, and predict patient outcomes. This information is crucial for treatment planning and determining the appropriate course of action (Liao et al., 2023; Go, 2022).

Digital pathology workflow optimization

AI can optimize the digital pathology workflow by automating manual tasks and streamlining processes. For instance, AI algorithms can facilitate slide organization, prioritize challenging cases for pathologists, and assist in quality control. This can enhance efficiency, reduce turnaround times, and alleviate the workload on pathologists, allowing them to focus on complex cases (Niazi et al., 2019).

Data-driven research and knowledge discovery

AI enables the analysis of large-scale pathology datasets, leading to new insights and discoveries. By mining pathology images and associated clinical data, AI algorithms can identify novel biomarkers, discover hidden patterns, and contribute to the understanding of disease mechanisms. This can accelerate medical research and foster innovations in diagnosis and treatment (Bohr & Memarzadeh, 2020; Quazi, 2022).

As with any AI application in healthcare, validation, and collaboration with experts in pathology are crucial to ensure the accuracy, reliability, and clinical relevance of AI algorithms. Additionally, addressing ethical considerations, privacy concerns, and regulatory requirements are vital for the responsible integration and adoption of AI in pathology and histopathology practice.

Clinical decision support systems

Patient data analysis: CDSS collects and analyzes patient data from various sources, such as electronic health records (EHRs), laboratory results, medical imaging, and genetic information. AI algorithms process this data, identifying relevant patterns, trends, and risk factors associated with specific diseases or conditions.

Diagnostic support

AI-based CDSS can assist in the diagnostic process by analyzing patient symptoms, medical history, and test results. By comparing patient data with a vast knowledge base, AI algorithms can generate differential diagnoses and provide clinicians with a ranked list of potential diagnoses, helping to narrow down the possibilities and improve diagnostic accuracy (Sloane & Silva, 2020).

Treatment recommendations

CDSS can provide evidence-based treatment recommendations tailored to individual patients. By considering patient-specific factors, such as age, comorbidities, medication interactions, and genetic profiles, AI algorithms can suggest appropriate treatment options, dosages, and drug regimens. This helps clinicians select the most effective and personalized treatment plans (Mukhopadhyay et al., 2020).

Drug interactions and adverse event prevention

AI-powered CDSS can analyze medication lists and patient data to identify potential drug interactions, contraindications, and adverse events. It can alert healthcare providers to potential risks, helping them avoid harmful drug combinations or adverse reactions and ensuring patient safety (Choudhury & Asan, 2020).

Clinical guidelines adherence

CDSS can help healthcare providers adhere to evidence-based clinical guidelines and best practices. AI algorithms can compare patient data with established guidelines, alerting clinicians if any recommended interventions or screenings are missing or if there are deviations from standard protocols. This promotes standardized care and improves patient outcomes (Mahadevaiah et al., 2020).

Real-time alerts and reminders

AI-based CDSS can generate real-time alerts and reminders to healthcare providers. For example, it can notify clinicians of critical lab results, upcoming preventive screenings, or the need for specific follow-up actions. This helps prevent oversights, enhances communication, and ensures timely interventions (El-Kareh & Sittig, 2022).

Prognostic and predictive modelling

AI algorithms in CDSS can utilize patient data to generate prognostic and predictive models. By analyzing historical patient data and outcomes, AI can help predict disease progression, treatment response, and potential complications. This information aids clinicians in making informed decisions about prognosis, treatment planning, and risk stratification (Giordano et al., 2021).

It's important to note that CDSS powered by AI is designed to augment, not replace, healthcare professionals' expertise. It serves as a decision support tool, providing clinicians with additional information and recommendations to aid their clinical judgment. Collaboration between AI developers, clinicians, and domain experts is crucial for developing and validating AI algorithms, ensuring their accuracy, reliability, and clinical relevance.

Early disease detection and risk assessment

Early disease detection and risk assessment are areas where AI can play a significant role in healthcare. By analyzing large amounts of patient data and identifying patterns and risk factors, AI algorithms can help in identifying diseases at early stages and assessing an individual's risk of developing certain conditions. Here's how AI is utilized in early disease detection and risk assessment.

Data analysis and pattern recognition

AI algorithms can analyze diverse sets of patient data, including electronic health records, medical imaging, genetic information, lifestyle factors, and sensor data. By identifying patterns and correlations in this data, AI can recognize early indicators or risk factors associated with specific diseases (Bohr & Memarzadeh, 2020).

Risk stratification and personalized risk assessment

AI can help stratify individuals into different risk categories based on their unique characteristics and medical history. By considering multiple factors, such as genetic markers, biomarkers, demographics, and lifestyle factors, AI algorithms can provide personalized risk assessments. This enables targeted interventions, preventive measures, and screening protocols tailored to an individual's level of risk (Johnson et al., 2021; Shandhi & Dunn, 2022).

Predictive modeling and forecasting

AI algorithms can build predictive models that estimate the likelihood of developing certain diseases. By analyzing historical patient data and outcomes, AI can identify risk factors and develop algorithms that predict disease occurrence, progression, or treatment response. These models can assist in early detection and enable proactive interventions (Ghaffar Nia et al., 2023).

Image analysis for early detection

AI algorithms can analyze medical images, such as mammograms, CT scans, or retinal images, to detect early signs of diseases like cancer or retinopathy. AI-based image analysis can help identify subtle abnormalities or variations that may not be easily recognizable to human observers, improving the chances of early diagnosis (Bi et al., 2019).

Sensor data and wearable devices

AI-powered systems can analyze data from wearable devices and sensors to monitor individuals' health parameters and detect anomalies. For example, AI algorithms can analyze heart rate variability, sleep patterns, activity levels, and other physiological data to identify early warning signs of cardiovascular diseases, respiratory disorders, or other conditions (Huang et al., 2022; Sunny et al., 2022).

Integration with electronic health records

AI can be integrated into electronic health record systems to analyze longitudinal patient data and identify trends or deviations from normal patterns. By comparing current data with historical information, AI algorithms can help identify early signs of disease development or changes in health status (Davenport & Kalakota, 2019; Bajwa et al., 2021).

Population health management

AI can analyze population-level data to identify trends, risk factors, and disease patterns within communities. By identifying high-risk populations or geographic areas, healthcare resources can be allocated more effectively, and targeted interventions can be implemented to address specific health needs (Dash et al., 2019).

The use of AI in early disease detection and risk assessment holds significant potential for improving patient outcomes and reducing healthcare costs. However, it's important to ensure that AI algorithms are developed and validated using diverse and representative datasets to minimize biases and ensure equitable and accurate predictions. Collaborative efforts between AI researchers, clinicians, and public health experts are essential to develop robust and reliable AI models for early disease detection and risk assessment.

Remote monitoring and telemedicine

Remote monitoring and telemedicine have become increasingly important in healthcare, especially in situations where in-person visits are challenging or not feasible. AI plays a crucial role in enhancing remote monitoring and telemedicine capabilities, enabling continuous monitoring, efficient data analysis, and remote consultations. Here's how AI is utilized in remote monitoring and telemedicine.

Wearable devices and sensors

AI-powered wearable devices and sensors can continuously monitor various health parameters, such as heart rate, blood pressure, glucose levels, and activity levels. AI algorithms analyze the data collected from these devices to detect patterns, anomalies, or changes in health status. This enables early detection of health issues and facilitates proactive interventions (Manickam et al., 2022).

Remote data analysis

AI algorithms can process and analyze the vast amount of patient data generated through remote monitoring. By identifying trends, patterns, and outliers, AI can provide valuable insights into a patient's health condition. This analysis can help healthcare providers make informed decisions, monitor treatment effectiveness, and identify potential complications (Bajwa et al., 2021).

Real-time alerts and notifications

AI algorithms can generate real-time alerts and notifications based on the analyzed data from remote monitoring devices. When critical health parameters deviate from normal ranges or predefined thresholds, AI can alert healthcare providers, allowing them to intervene promptly and prevent adverse events (Dubey & Tiwari, 2023).

Teleconsultations and virtual care

AI can enhance teleconsultations by providing decision support to healthcare providers. AI algorithms can analyze patient data, medical records, and symptoms to assist clinicians in making accurate diagnoses and treatment recommendations during virtual appointments. AI-powered chatbots can also offer personalized health advice and answer patient queries, improving access to healthcare information (Davenport & Kalakota, 2019).

Natural Language Processing (NLP)

NLP techniques combined with AI enable automated analysis of patient-generated health data, such as electronic health records, medical notes, and patient-reported symptoms. NLP algorithms can extract relevant information, identify key clinical concepts, and provide summaries or insights to support remote consultations and streamline the documentation process (Alzoubi et al., 2019).

Remote imaging interpretation

AI algorithms can assist in the interpretation of medical images, such as X-rays, CT scans, or dermatological images, remotely. By analyzing these images, AI can detect abnormalities, assist in diagnosing conditions, and provide preliminary findings to healthcare providers. This allows for faster and more efficient diagnosis, even in remote settings (Aggarwal et al., 2021).

Remote triaging and prioritization

AI algorithms can help prioritize patients based on the severity of their conditions during remote consultations or when accessing healthcare remotely. By analyzing patient data, symptoms, and risk factors, AI can assist in triaging patients, ensuring that those in urgent need of care receive prompt attention and appropriate follow-up (Villafuerte et al., 2023; Nazir et al., 2022; Sarker et al., 2021).

The integration of AI into remote monitoring and telemedicine offers opportunities for enhanced healthcare access, reduced costs, and improved patient outcomes. However, it is crucial to address concerns related to data security, privacy, and ethical considerations when deploying AI-powered systems in remote healthcare settings. Additionally, it is important to maintain a balance between remote care and in-person care to ensure comprehensive and appropriate patient management.

Conclusion

The future of AI in disease diagnosis holds great potential. As AI algorithms continue to improve and more healthcare data becomes available, we can expect even more accurate and efficient diagnostic tools. AI could facilitate the development of personalized medicine, where treatments are tailored to an individual's unique characteristics, improving patient outcomes and reducing healthcare costs. Ethical considerations, privacy concerns, and regulatory frameworks will also need to be carefully addressed to ensure the responsible and equitable use of AI in healthcare.

It's important to note that while AI can be a powerful tool in disease diagnosis, it is not meant to replace healthcare professionals. AI systems should be seen as decision-support tools that augment human expertise, assisting clinicians in making more accurate diagnoses and improving patient care.

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Conflict of interest

All authors declare that there is no conflict of interest in this work.

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