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Artificial Intelligence in the Diagnosis of Disease: An Analytical Review on the Current Trend in Research Leading to Better Outcomes

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ABSTRACT

Artificial intelligence (AI) has profoundly influenced various industries, including healthcare. AI is revolutionizing medical diagnostics by significantly improving the efficiency and accuracy of disease diagnosis. This study examines the current trends of AI in disease diagnosis, focusing on its potential to utilize vast clinical data and genomics to enhance clinical predictions, disease diagnosis, and prevention, ultimately leading to better patient outcomes. This study aims to examine the latest trends in AI applications in disease diagnosis. It helps to understand how these advances can transform healthcare. This analytical review is based on an extensive literature search across academic databases such as Scopus, PubMed, Web of Science, and Elsevier using inclusion/exclusion criteria focused on AI techniques and their applications in disease diagnosis. The findings highlight the rapid growth of AI in medical diagnostics, showcasing its ability to process and analyze extensive patient data efficiently to make accurate and precise diagnosis of disease. The continuous evolution of AI technologies enables the rapid evaluation of extensive patient data, including medical history, demographics, and laboratory test results. This advancement makes diagnostics more predictive, preventative, and precise, promising a transformation in healthcare. However, it is essential to recognize that although AI can be a powerful tool, it cannot replace the clinician's role in disease diagnosis.

Keywords: Artificial intelligence, Diagnosis, Disease, Diagnostics

Introduction

Diagnosis is the process of identifying a disease by evaluating symptoms, medical history, and diagnostic tests. Diagnostic methods including blood tests, histopathology, biopsy procedures, medical imaging, bio-signals, and genomics are crucial in identifying the underlying causes of medical issues.[1,2] The outcomes of these diagnostic tests enable clinicians to devise optimal treatment plans for their patients, track the progression of a condition or disease, and gauge the effectiveness of treatment (prognosis). Diagnostics is a highly complex process.[2,3] However, diagnostics is critical to ensure proper diagnosis of disease for efficacious treatment and effective patient care.[2,4,5]

The use of artificial intelligence (AI) as an element of the diagnostic process has been steadily growing.[2,5–7] Healthcare professionals have been using it from numerous perspectives.[8–12] There is no consistent definition for the term AI.[13] However, it may be considered as "the ability of a machine to perform cognitive functions that we associate with human minds, such as perceiving, reasoning, learning, interacting with the environment, problem-solving, decision-making, and even demonstrating creativity" and is generally analogous to human-like behavior with enormous applications.[14–16] AI can rapidly process vast amounts of patient data, encompassing imaging, bio-signals, vital signs (such as body temperature, pulse rate, respiration rate, and blood pressure), demographic details, medical history, and laboratory test results.[17–20] Nevertheless, the utilization of AI in healthcare and disease diagnostics is generally focused.[3,21,22] It is being developed using machine learning.[23–26]

Algorithms are used in medical data to generate predictions.[27,28] The data are continually gathered from various sources and updated over time by processing new and relevant information.[29–31] AI-empowered systems can process more information at a faster pace compared with humans and may outperform them for certain medical tasks.[3,31] Incorporating AI into diagnostics can expedite the identification of pertinent medical data from multiple sources related to the patient and treatment process.[27–31] The diversity of patient data in terms of multimodal inputs can lead to better diagnostic decisions based on multiple findings from images, signals, and text representation.[13,14] By integrating these multiple data sources, healthcare providers can achieve a more comprehensive understanding of a patient's health and the underlying causes of their symptoms. This multimodal data can offer a complete picture of a patient's health, reducing the risk of misdiagnosis and enhancing diagnostic accuracy.[13,27,31]

We have explored the general application of AI in healthcare and delved into the current state of AI in disease diagnostics.[14,32–36] The determination of a disease diagnosis involves assessing whether a patient is affected by a specific condition, which can vary based on individual experiences and emotional and environmental factors.[37–40] Diagnosis can be envisioned as a "pre-existing set of categories agreed upon by the medical profession to designate a specific condition."[39,40] Diagnosing typically requires the expertise of professionals from various medical disciplines.[41,42] Therefore, more healthcare professionals rely on information technology.[3,13,43,44] One of the most significant areas where AI has made an impact is medical imaging techniques such as CT scans, X-rays, ultrasound, and MRI which generate vast amounts of data that can be challenging for human radiologists to analyze thoroughly.[3,45–48]

AI algorithms, particularly those based on deep learning, have demonstrated remarkable proficiency in interpreting medical images. AI algorithms, especially those utilizing deep learning, have shown exceptional skill in interpreting medical images.[3,6] AI is already enhancing disease diagnosis, such as in the early detection of ectopic pregnancies and aiding gynecologists in making early treatment decisions.[38,49] The role of AI in cancer diagnosis, cardiovascular disease, lung disease, histopathology, dermatology, and genomics has also been substantial.[18,50–53]

Methods

The review article is based on an extensive literature search in academic databases such as Scopus, Google Scholar, IEEE Xplore, PubMed, Web of Science Springer, and Elsevier. It employs specific inclusion/exclusion criteria, focusing on data extraction related to AI techniques and their applications in diagnosis.

Results

A comprehensive analysis of the collected information was conducted to understand AI's role in diagnosing various diseases. Figure 1A and B, generated using the GraphPad Prism 10 software, provides an overview of the literature on medical diagnosis, patient classification, and prognosis.

Figure 1A and B presents the results of the literature overview on diagnosis, prognosis, and patient risk stratification over the past five years from various databases. The Fig 1A shows a bar plot depicting the number of articles identified per search query for each database. The Fig 1B illustrates the timeline of the total number of articles published per year, aggregated from the various databases, created using the Graph Pad Prism 10 software.

Discussion

The acquisition and evolution of techniques used in AI have taken place due to upgradation in the technology of processors by increasing speed and storage capacity.[7,12,54–56] It has resulted in rapidly acquiring and processing information from different sources at a lower cost.[43,45] The accessibility of techniques is easy and hence can be used effortlessly.[45,47] The commonly used techniques are generally classified as supervised, unsupervised, and deep learning.[6,14,29,53] The algorithms employed by using existing samples or data in the case of supervised learning and association are based on labels of a dataset that are known. Images of fractures or ruptures that have been classified by medical specialists may be considered as an example of supervised learning. In turn, the information is used to instruct algorithms that will generate predictions for unused data.[13,27,28] It is dependent on the user's input and is highly sensitive to data quality.[29–31] Nowadays, supervised learning is one of the most commonly used approaches and gives sturdy classifications. Examples of supervised learning approaches have been used in the diagnosis of dementia and cancer.[31,37,53,57,61]

Table 1 lists the key role played by AI in medical diagnostics. There is another technique, which is known as unsupervised learning that utilizes self-organizing algorithms without existing samples or training data.[6,14,52] For example, unsupervised learning is used in hepatitis diagnostics.[58] Another AI technique that is routinely used is deep learning, which is an individual AI approach; however, it has the capability of combining both supervised and unsupervised approaches.[3] The classification of dermatological diseases and atrial fibrillation detection are examples of deep learning.[29,57,60,62]

There are challenges in the operation of AI technologies in healthcare systems, even though this is one of the most critical advancements in biomedical research.[3,7,32,35] In ever-evolving AI techniques such as the Boltzmann machine, K-nearest neighbor, support vector machine (SVM), decision tree, logistic regression, fuzzy logic, and artificial neural network to diagnose diseases are being used with remarkable accuracies.[47,57–60]

For example, a hybrid intelligence system was used in diagnosing skin disease to achieve the highest level of accuracy.[29,57] In another study, a recurrent neural network was used to diagnose hepatitis virus and achieved 97.59% accuracy, while a feed-forward neural network achieved 100%.[58] One research finding in gastroenterology got a 97.057 area under the curve by using residual neural network and long short-term

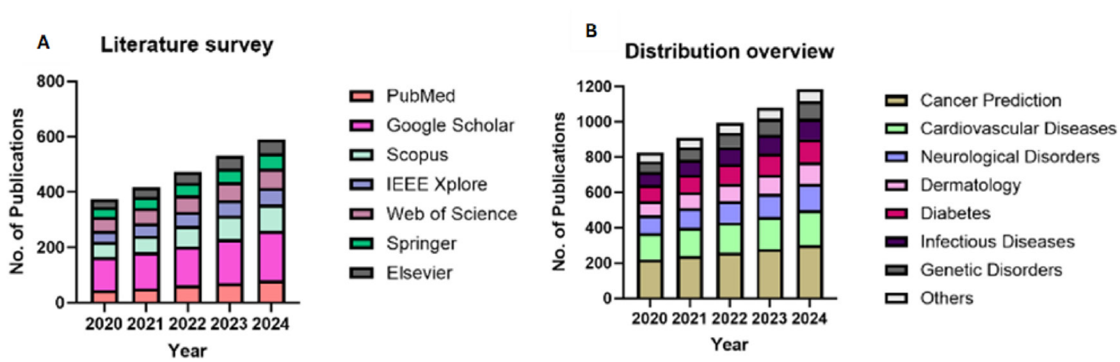

Fig 1 | (A) Distribution of published papers over the past five years. (B) Distribution published papers on various categories of disease diagnosis using artificial intelligence

Table 1 | Encompasses AI in Medical Diagnosis and their Attributes

AI Role	Disease Area	Description
Machine Learning Models	Cancer	Predicting cancer types and stages using patient data
Deep Learning Algorithms	Radiology	Analyzing medical images for disease detection
Natural Language Processing (NLP)	Medical Records	Extracting relevant information from electronic health records (EHRs)
Predictive Analytics	Cardiology	Predicting cardiovascular diseases based on patient history
AI-based Diagnostic Tools	Dermatology	Detecting skin cancer through image analysis
Reinforcement Learning	Personalized Medicine	Optimizing treatment plans for individual patients
Genomic Data Analysis	Genetic Disorders	Identifying genetic mutations linked to various diseases
Speech Recognition	Neurology	Diagnosing neurological disorders through speech pattern analysis
Predictive Modeling	Infectious Diseases	Predicting outbreak and spread of infectious diseases
AI-enhanced Pathology	Pathology	Assisting pathologists in analyzing tissue samples

memory (LSTM) to diagnose gastrointestinal disease.[50] An interesting study showcased a self-determining dual-analysis examination by engaging ladies of 50–69 years and mammography.[61] Accumulation of the data-building tool resulted in a non-significant enhancement in sensitivity by 76.2% and a significant increase by 96.4%. The random forest classifier stood out to be the best algorithm in Type 2 diabetes diagnosis in large groups.[28,30]

In a study on predictive coronary heart disease using medical data with the help of three supervised learning techniques, namely, Naïve Bayes, SVM, and decision tree, on the South African Heart Disease dataset of 462 instances to find the correlations in coronary heart disease, which would help improve the prediction rate reported ten-fold cross-validation.[29,63] In a study that used Internet of Things (IoT) for a healthcare monitoring system for diabetes and hypertension patients at home and used personal healthcare devices that perceive and estimate a person's biomedical signals. The system can notify health personnel in real time when patients experience emergencies.[3,14] They applied the administered AI method to achieve parallel grouping from the 18th lower request shading minutes. Their test indicated a precision of 98.4%, particularly for the tuberculosis antigen explicit counteracting agent identification on the portable stage.[64] A study provided the global trends and developments of AI applications related to stroke and heart diseases to identify the research gaps and suggest future research directions.[29,63] The research proposed an effective model that can help doctors diagnose skin diseases efficiently that combined neural networks with MobileNet V2 and LSTM and achieved an accuracy rate of 85%, exceeding accuracy and generating faster results as compared to the traditional methods.[3,6,14,24] The cervical cancer prediction model for early prediction of cervical cancer using risk factors as inputs using a combination of AI techniques showed better accuracy than previously proposed methods for forecasting cervical cancer.[31,35,57]

The advancement in AI in diagnostics over the conventional approach is shown 100× faster than conventional methods (Figure 2, created by BioRender). This illustrates the progression of AI in medicine, particularly in enhancing healthcare from bench to bedside.

Figure 2 illustrates the progression of AI in medicine, particularly in enhancing healthcare from bench to bedside. Patient-derived datasets are employed in AI models to accelerate this transition. Integrating datasets from multiple diverse sources, including sequencing, structural, and screening data, expands the relevant feature space for AI models, facilitating comprehensive end-to-end medical discovery.

Figure 3 depicts the progression and application of AI in healthcare research showcasing the following highlights:

Research Timeline: Depicts the stages of research from basic research, which includes predicting protein structure and automating high-throughput assays, to clinical research that integrates multimodal data for improved patient outcomes.

Standard Image Analysis: Shows the use of off-the-shelf tools such as 3D Slicer, Qu Path, Fiji, Napari, and ilastik for different scales of biological imaging from whole body to subcellular levels.

Building a Pipeline: Describes the iterative process of training, tuning, testing, validating, and deploying AI models using large datasets for repetitive tasks.

Foundation Models: Explains the application of foundation models, including supervised fine-tuning for few-shot learning and direct application for zero-shot learning, in solving complex problems. As far as the current trends in the research, it may be said that there is a profound and multifaceted role of AI in medical diagnostics, which has been significantly noticeable in the field of medical imaging and radiology, providing tools that assist in the accurate and rapid diagnosis of various conditions (Figure 4). DL models, especially convolutional neural networks, have become necessary in analyzing medical images.[3,6,18] AI techniques and models have shown exceptional performance in diagnosing lung diseases such as pneumonia, tuberculosis, and lung cancers.[65–68] AI's role in mammography has improved breast cancer detection rates by reducing false positives and negatives.[61] Studies have demonstrated that AI algorithms can surpass the diagnostic accuracy of radiologists. AI algorithms have been shown to accurately identify and classify brain tumors from scans, helping in early diagnosis and treatment planning. In pathology, AI aids in analyzing histopathological images, improving diagnostic precision and efficiency.

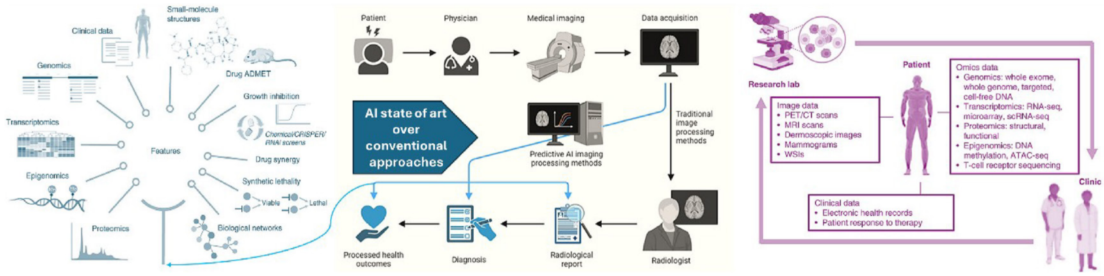

Fig 2 | Advancement of AI in diagnostics over conventional approaches (BioRender.com/q62k409)

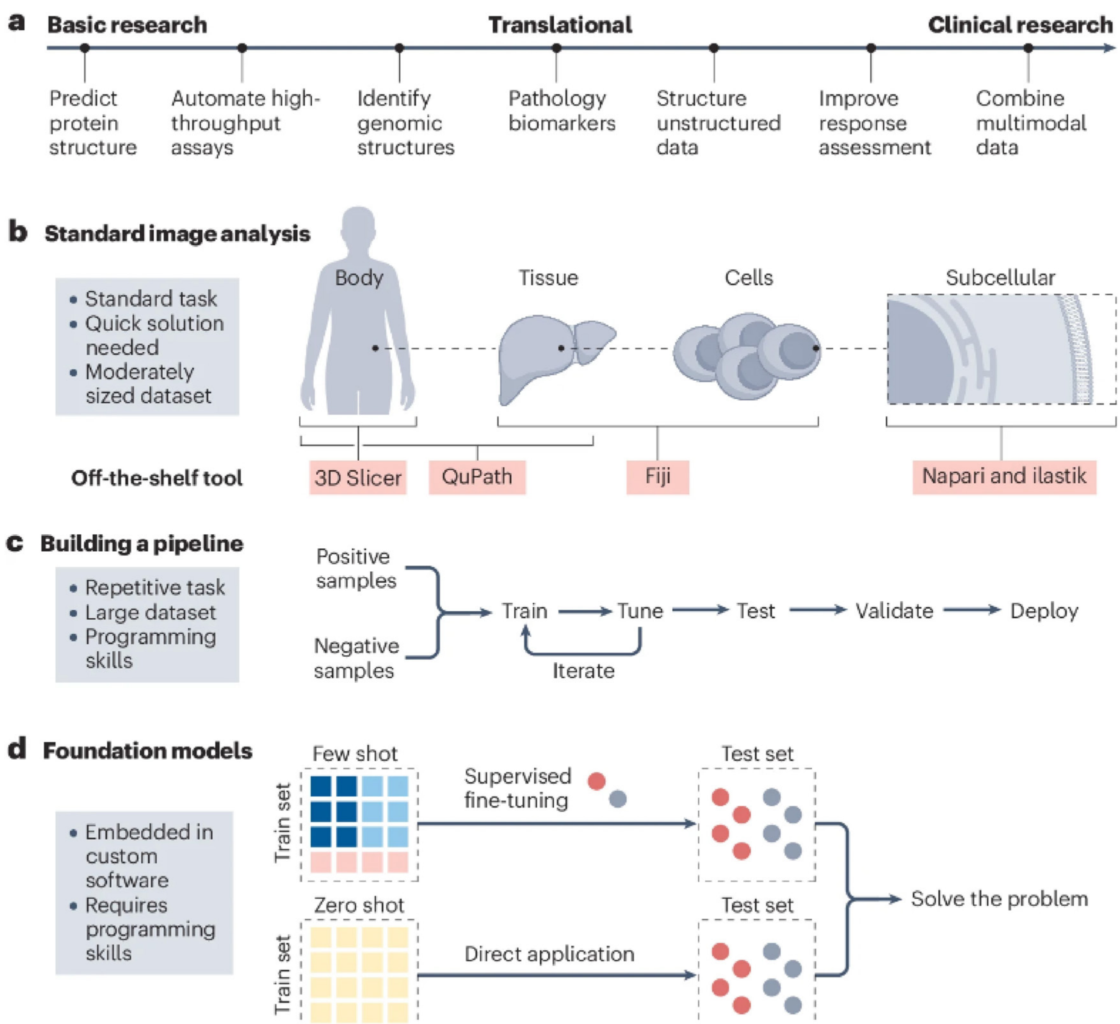

Fig 3 | State-of-art advancements in AI for medical research and applications. Adapted from Perez-Lopez et al.[53]

AI algorithms can identify and classify cancerous cells with high accuracy, which is crucial for effective treatment planning. AI models have been developed to accurately classify prostate cancer from biopsy images, assisting in early detection and personalized treatment plans.[18] AI has improved the accuracy of breast cancer diagnosis by detecting morphological features in tissue samples.[31,37,53,61] Apart from the scope of the present review, which was focused on diagnostics, it may be added that AI plays a pivotal role in genomics by analyzing vast amounts of genetic data to identify markers and predict disease susceptibility. AI models can process complex genomic datasets, leading to breakthroughs in personalized medicine.[18,57] AI models predict patient outcomes based on historical data, guiding treatment decisions and improving patient care. Furthermore, AI has revolutionized drug discoveries, predicting their efficacy and safety profiles.

This has accelerated the drug development process, reducing the time and costs associated with traditional methods. AI technologies enable continuous monitoring of patients providing early warnings and

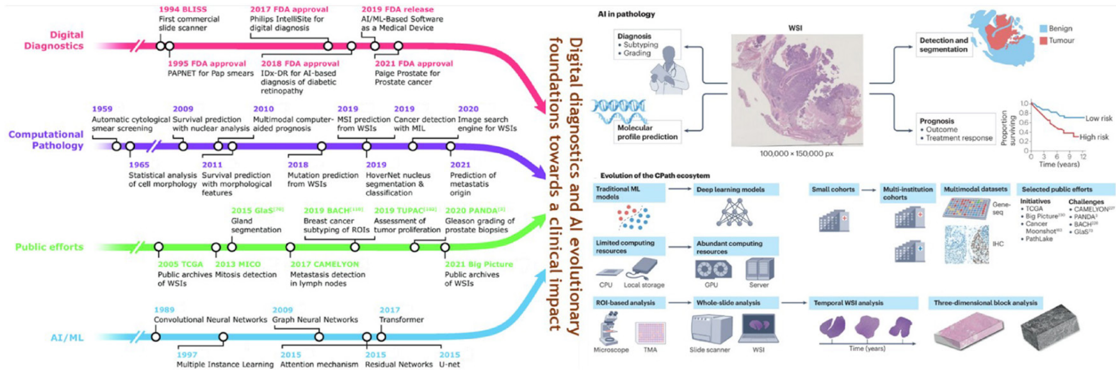

Fig 4 | AI workflows in medical prediction and diagnosis: Applications, timeline of selected milestones, and trend in clinical impact

improving chronic disease management. AI-powered wearable devices monitor vital signs and detect anomalies, alerting healthcare providers to potential health issues.[3,7] AI deployment in medical diagnostics is still in the early stages, and several technical, regulatory, and ethical challenges must be overcome to reach their full potential.[12,32,35] The first challenge is medical data quality and availability. Meanwhile, AI algorithms can be biased if that is not representative of the population they are intended to serve, leading to incorrect or unfair diagnoses. Another aspect is the use of AI in medical diagnostics of private and sensitive data, which raises some ethical questions. To overcome these ethical issues, ongoing deliberations and federated learning are being explored as potential solutions.

The review presents a novelty by offering a comprehensive literature survey that highlights various disease diagnoses using emerging AI techniques, distinguishing it from most literature reviews that typically focus on just one or two diseases. Additionally, it emphasizes how different AI techniques in research publications have predominantly concentrated on the accuracy of the methods employed, which is a critical criterion for diagnosing specific illnesses. Furthermore, this review provides insights into future research directions for employing specific AI techniques aimed at the early diagnosis and management of chronic, debilitating diseases and disorders and enhancing quality of life by reducing medical costs and burdens.

Conclusion

The future of AI in medical therapeutics and diagnostics is promising, with ongoing research focused on integrating AI with other emerging technologies. Combining AI with the IoT can enhance remote patient monitoring and data collection, leading to more comprehensive health insights.

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