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Original Article

Disease Diagnostic Systems based on AI-Applications in Healthcare: Models, Challenges, and Future Directions

Vandana Chaturvedi
Independent Researcher, India.

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Abstract *- Artificial Intelligence (AI) is improving clinical decision-making, accuracy, and efficiency, which is revolutionizing disease diagnosis systems. AI-powered diagnostic technologies can now evaluate medical images, including magnetic resonance imaging, computed tomography, and radiographs, allowing doctors to identify patients more rapidly and precisely. AI technologies, such as machine learning (ML), deep learning (DL), and natural language processing (NLP), are being used to handle both organized and unstructured healthcare data at a rapid pace. It has been shown that Decision Tree (DT), Support Vector Machines (SVM), K-Nearest Neighbors (KNN), Naive Bayes (NB), Logistic Regression (LR), and AdaBoost algorithms may successfully analyze clinical parameters of illnesses, including diabetes, renal disease, and heart disease. Time-series analysis, signal processing, and medical imaging all make extensive use of Convolutional Neural Networks (CNNs) and Recurrent Neural Networks (RNNs), two key models in the DL paradigm. Medical imaging, cardiovascular prediction, chronic disease management, neurological assessment, infectious disease detection, and precision medicine are among the applications supported by AI-based systems. Although the creation of AI in healthcare is associated with issues regarding data quality, standardization, interpretability, bias, and compliance with regulations, the future of AI is marked by progress in robotics, oncology, digital pathology, genomics, and integration of AI with the IoT. AI-based diagnostic tools are anticipated to enhance the current state of healthcare delivery by increasing accuracy, efficiency, and accessibility.*

Keywords *- Artificial Intelligence (AI), Natural Language Processing (NLP), Disease Diagnostics, Medical Imaging, Precision Medicine and Healthcare Applications.*

1. Introduction

Diagnostic Disease in Healthcare is a very basic application of establishing a disease or a medical condition that has clarified the symptoms and the clinical manifestations of a patient. It involves systematic measurement through medical history, physical examination and in case of necessity, laboratory measurements to make effective clinical judgements. In medical disciplines, medical diagnosis establishes a disease or diseases, which clarify the symptoms and signs of an individual. It is the history of the patient and his physical examination that often helps in gathering the data on diagnosis[1]. As a result, developing countries such as Bangladesh and India are faced with the dilemma of providing satisfactory diagnostic processes to the largest number of patients since they do not have enough health professionals to serve their population[2]. The first endeavors in the medical sector were associated with expert systems that tried to replicate the human thought processes in the area of cognitive thinking [3]. The first improvement efforts in healthcare were primarily aimed at improving clinical decision-making through the aid of a properly structured method of computations. The active work on the digitalization of healthcare technologies, the notion of AI takes center stage in the modern disease diagnostics system, enabling a person to conduct clinical assessment faster, more accurately, and on a data-driven basis.

The integration of ML and DL algorithms has allowed automated pattern recognition, feature extraction, and predictive modeling of medical data across a variety of medical data[4]. The technologies work best in areas where medical imaging, signal processing, and mass clinical data analysis are used to diagnose an individual[5][6]. AI-based diagnostic models can help decrease the level of human error, enhance clinical outcomes, and allow longitudinal tracking of patient health records. The foundation of AI-based disease detection is ML, which is widely classified into three categories: Reinforcement learning, supervised learning, and unsupervised learning[7][8]. Disease categorization is another job carried out by supervised learning models, and DL models, especially CNN, have proven to have higher performance results in medical image analysis because they are capable of automatically producing high-level features[9].

Despite these developments, there are still barriers to broad clinical usage, like lack of standardization, heterogeneity of data, interpretability issues and regulatory impediments[10][11]. Consequently, knowledge of AI models and their uses in healthcare, the level of challenges, and future development direction is required to promote reliable and scalable disease diagnostic systems. In this paper, a systematic overview of the AI-based disease diagnostic systems is provided with special focus on the underlying models, clinical applications, implementation issues, and future research perspectives in the field of intelligent healthcare diagnostics.

1.1. Organization of the Paper

The remainder of this paper is structured as follows: Section II discusses AI in disease diagnostics, including ML, DL, and NLP techniques. Section III presents applications in medical imaging, chronic and cardiovascular disease prediction, neurological and infectious disease detection, and precision medicine. Section IV outlines implementation challenges and future directions, and Section V highlights the summary of the review. Finally, Section VI concludes with key findings and insights on AI-driven advances in healthcare.

2. AI in Disease Diagnostic Systems

The use of AI for medical diagnosis is still in its infancy, but the availability of more data is opening the door to extensive applications, particularly in the detection of disorders like cancer. The intricacy of the underlying mechanisms and symptoms in the particular cases of infectious diseases and sepsis makes the development of early diagnostic techniques difficult. Nevertheless, ML models have potential for diagnosis through task automation, workflow management, and decision making.

2.1. AI Technologies in Healthcare

Healthcare AI is an intersection of computer systems, which are created to simulate the behavior of a human brain, including learning, reasoning, and problem-solving. This chapter explores different AI healthcare technologies (see Figure 1).

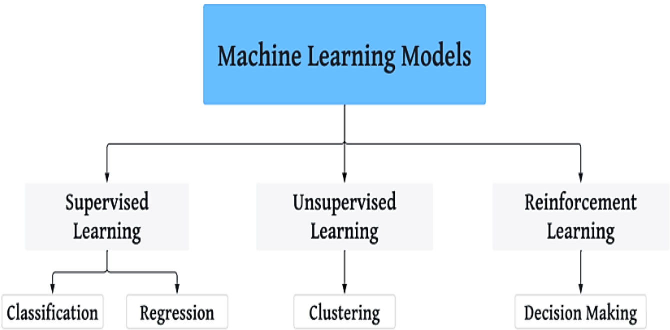

Fig 1: Machine Learning Models and Algorithms

2.1.1. ML in Healthcare

ML is an essential part of AI because it employs statistical tools to interpret and analyze large volumes of data, which enables breakthroughs in healthcare. This section provides the most frequently used ML algorithms in disease diagnosis.

- Decision Tree (DT): The divide-and-conquer strategy is used by the DT algorithm. The attribute may have different values in DT models, which are referred to as classification trees; branches display the set of traits that lead to the classifications of classes, while leaves indicate various classes. Conversely, DT may use regression trees, which are continuous variables. The two most widely utilized and well-liked DT algorithms are C4.5 and EC4.5.
- Support Vector Machine (SVM): A common ML method for issues requiring regression and classification is the SVM. Text classification is one of the many fields in which SVMs have been widely used, in addition to disease diagnosis, distant homology identification, protein fold, voice recognition, and facial expression recognition. Supervised ML methods cannot handle unlabelled data. SVM can categorise unlabelled data by identifying groupings within the data using a hyperplane.
- K-Nearest Neighbor (KNN): In disease detection systems, KNN is a non-parametric supervised learning approach for applications involving regression and categorisation[12]. It categorizes the state of a patient by the most similar cases of patients in the dataset and places the patient in the majority of them. Medical records are typically compared using distance measures like Euclidean distance. In regression-based diagnostic use cases, including predicting the severity or risk factors of the disease, it is calculated as the average (or weighted average) of the samples that are most similar to one another.
- Naïve Bayes (NB): The NB classifier presumes that features are conditionally independent and is a probabilistic ML technique grounded in Bayes' theorem. With the help of the observed feature values, the NB classifier. compute the posterior probability of each of the possible class labels, given a data instance. The case is then classified under the most probable class. Instead of simply giving deterministic predictions, the Naive Bayes classifier gives an estimation of the probability and posterior probabilities of the class membership. It is widely used for classifying large-dimensional datasets, where it is well represented by its simplicity, computational efficiency, and performance on high-dimensional datasets for medical diagnosis, text classification, and disease prediction.
- Logistic Regression (LR): An ML technique called LR is applied to classification issues. With predicted values between 0 and 1, the LR model has a probabilistic structure. Some examples of LR-based ML include identifying fraudulent online transactions, detecting spam emails, and diagnosing malignant tumours. LR employs the sigmoid function that is occasionally called the cost function. The sigmoid function converts all real numbers from 0 to 1.
- AdaBoost: To generate a powerful prediction model, the ensemble learning method AdaBoost (Adaptive Boosting) combines several weak learners. It works by giving samples that were incorrectly classified a greater weight and samples that were correctly classified a lower weight, enabling subsequent learners to focus on harder cases. AdaBoost may be used for tasks involving both regression and classification, improving overall model accuracy and robustness.

2.1.2. Supervised Learning

The input data and the appropriate output are matched in labelled datasets used to train the models. Supervised learning is commonly used for tasks like regression (e.g., patient outcome prediction) and classification (e.g., disease diagnosis). For instance, using patient demographics and medical history, heart disease risk has been predicted using supervised learning algorithms.

2.1.3. Unsupervised Learning

Unsupervised learning models identify groupings and patterns in unlabelled data, in contrast to supervised learning[13]. These models are very helpful for identifying irregularities in medical data or grouping individuals with similar traits. As an illustration, to design each patient's unique therapy, unsupervised learning was used to group patients by their genetic profiles.

2.1.4. Reinforcement Learning

Reinforcement learning is an ML methodology whereby the models are trained to make sequential decisions through engaging with the environment and being rewarded or punished corresponding to their actions. The objective is to obtain a better strategy that optimizes long-term cumulative benefits. Reinforcement learning has demonstrated potential in healthcare to enhance treatment strategies, such as personalizing medical regimens and determining the optimal chemotherapy dose for cancer patients.

2.2. DL in Healthcare

To handle complex data and generate precise results, DL, a sophisticated form of ML, employs multilayer neural networks. Key DL methods include:

2.2.1. Convolutional Neural Networks (Cnns):

CNNs are tremendously useful in medical image processing because they are well-suited for image analysis. Convolutional layers are used by these networks to identify features and image patterns, like cancers in radiological scans[14]. Specifically, CNNs have shown successful in detecting diabetic retinopathy in retinal images and breast cancer in mammograms.

2.2.2. Recurrent Neural Networks (Rnns):

RNNs are designed to process sequential data, such as time series and text. RNNs have been applied in healthcare to predict patient outcomes using time-series data from wearable technology and electronic health records (EHRs), enabling early identification of health problems.

2.3. Natural Language Processing (NLP) In Healthcare

The medical industry depends heavily on NLP, a subfield of AI that examines and deciphers human language. NLP techniques include:

- Text classification: Research articles, clinical notes, and patient data may all be classified by NLP algorithms into predetermined categories. For instance, the triage procedure in emergency rooms has been enhanced by the automated classification of patient concerns using NLP.
- Named entity recognition (NER): In unstructured text, NER algorithms have the ability to identify certain data, like the patient's name, diagnosis, and medication. This is required for the automated extraction of pertinent data from EHRs.
- Sentiment analysis: NLP can be used to evaluate the opinion of the population towards the healthcare services, potential interventions to make a better patient treatment, through examining social media posts and patient comments.

2.4. The role of AI in Symptom-Based Disease Diagnosis

The AI plays a critical role in disease diagnosis by using symptoms in Smart symptom-checking chatbots, which provide free, immediate results from the first health tests. Such systems can be employed to process the symptoms entered by the user through the use of NLP, ML and structured medical knowledge to facilitate the early intervention, promote prompt intervention and allow the patient to participate in active health management practices[15].

In the presented comparative analysis, Table I shows the application of the technologies of ML and DL in the work with different diseases, including kidney disease, cancer, diabetes, and heart diseases[16]. The review suggests that ML techniques are primarily effective when working with structured clinical information, and DL models exhibit strong potential for handling complex diagnostic procedures involving imaging and signal types.

Table 1: Machine Learning vs Deep Learning in Disease Diagnostics

Approach	Disease	Clinical Indicators	Methods Used	Research Objectives
Machine Learning	Kidney Disease (CKD)	Edema, hypertension, abnormal creatinine, reduced GFR	SVM, KNN	Comparative performance evaluation
	Kidney Disease	Proteinuria, hematuria, electrolyte imbalance	KNN, SVM, Decision Tree, Logistic Regression	Support clinical treatment decision-making
	Breast Cancer	Breast lump, nipple discharge, abnormal mammogram findings	K-Means + SVM (K-SVM)	Tumor classification (WDBC dataset)
	Breast Cancer	Tumor size, texture irregularities, imaging abnormalities	SVM, Decision Tree, Naïve Bayes, KNN	Comparative algorithm analysis

	Arthritis	Joint stiffness, swelling, inflammation	CART with WEKA	Predict disease progression and patient improvement
	Diabetes	Hyperglycemia, fatigue, polyuria, blurred vision	SVM	Early detection of Diabetes Mellitus
	Diabetes	Blood glucose variation, slow wound healing	KNN, SVM, Random Forest, J48	Comparative performance analysis
	Parkinson's Disease	Tremors, speech impairment, bradykinesia	SVM, KNN	Voice signal-based diagnosis
	Parkinson's Disease	Balance disorders, muscle rigidity	SVM	Differentiation between PD and PSP patients
	Influenza	Fever, cough, sore throat, body ache	ML Classifier + Fuzzy Logic	Reduce uncertainty in detection
	Liver Cancer	Jaundice, abdominal pain, liver enlargement	Neural Network, Fuzzy C-Means, Bayesian Models	Detection of normal vs abnormal regions
Deep Learning	Skin Disease	Rashes, lesions, pigmentation changes	Deep CNN (DCNN)	Automated skin disease detection
	Skin Cancer	Asymmetrical moles, irregular borders, color variation	CNN + Multiclass SVM	Early melanoma detection
	Breast Cancer	Clinical Indicators Considered	Deep Belief Network + Backpropagation	CAD-based tumor diagnosis
	Breast Cancer	Edema, hypertension, abnormal creatinine, reduced GFR	K-Means + Image Processing	Tumor region segmentation
	Diabetes	Proteinuria, hematuria, electrolyte imbalance	CNN, LSTM (ECG/HRV signals)	Signal-based diabetes classification
	Diabetes	Breast lump, nipple discharge, abnormal mammogram findings	Decision Tree, SVM, Naïve Bayes	Diabetes prognosis prediction
	Heart Disease	Tumor size, texture irregularities, imaging abnormalities	Deep CNN	Automated heart sound classification
	Heart Disease	Joint stiffness, swelling, inflammation	DNN	Diagnosis and prognosis support
	Lung Cancer	Hyperglycemia, fatigue, polyuria, blurred vision	CNN, Deep Belief Network (DBN), Stacked Denoising Autoencoder	Lung cancer detection (LIDC dataset)

3. Applications of AI-Based Disease Diagnostic Systems

The use of AI in healthcare may be found in a variety of context, which might all improve the quality of patient treatment, advance medication research, and foresee outbreaks.

3.1. Medical Imaging-Based Diagnosis

AI-assisted systems in disease diagnostics have transformed medical imaging by making it easier to identify diseases early, perform proper classification, and optimize treatment using DL algorithms. CNNs, U-Net, GANs, and Image segmentation may be enhanced by using transfer learning models, reconstruction, and lesion detection in MRI, CT, PET, and mammography[17]. Multimodal image fusion can further enhance the accuracy of tumor localization and staging by integrating anatomical and functional information. Altogether, the AI-based imaging system is moving towards automated, precision-oriented clinical decision support.

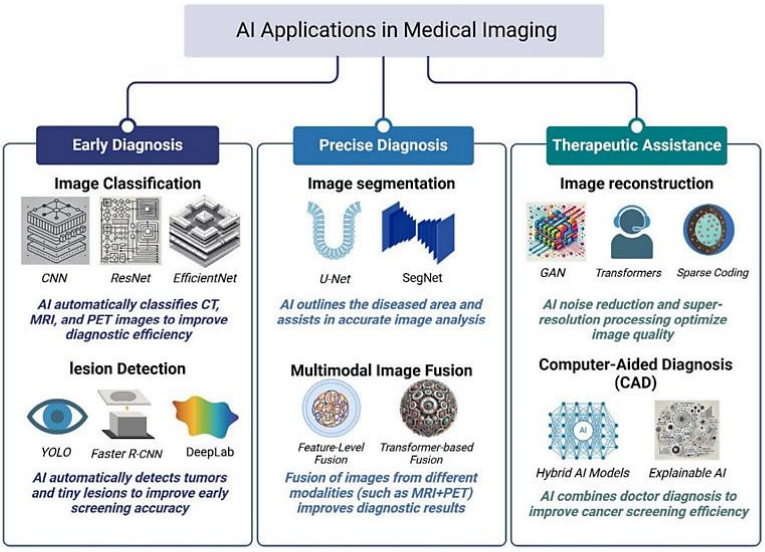

Fig 2: AI Applications in Medical Imaging

Figure 2 highlights the three major categories of AI's primary applications in medical imaging: early diagnosis, Precise diagnosis, and Therapeutic support. (1) Early Diagnosis: Image classification and lesion detection, which makes it possible to screen cancer automatically and identify abnormalities in medical images (e.g., mammography, CT, MRI, PET). (2) Precise Diagnosis: include multimodal image fusion, image separation, enhancing tumour localization, drawing borders, and raising diagnostic precision. (3) Therapeutic Support: Takes image reconstruction and computer-aided diagnosis (CAD), which is used to improve the quality of the image and aid in clinical decision-making. Each application uses certain image data formats and DL techniques (e.g., CNN, U-Net, GAN).

3.2. Cardiovascular Disease Prediction

AI-based diagnostic tools can forecast cardiovascular diseases because of the advanced processing of cardiac sounds, ECG signals, retinal images, and cardiac CT scans. The deep learning models and especially CNN-based systems are able to detect arrhythmias, heart failure, valvular disorders, and coronary artery disease at a higher accuracy and speed rate in comparison to the traditional approach[18]. Multimodal integration based on AI allows adjusting risks on an individual basis and developing treatment plans accurately[19]. All in all, the technologies help in earlier intervention, improved patient outcomes and effective cardiovascular care management.

3.3. Diabetes and Chronic Disease Prediction

The early detection and treatment of diabetes and other chronic conditions, including cardiovascular disorders and hypertension, are some of the critical aspects of AI-based diagnostic systems. ML and DL algorithms examine electronic health records, lab results, lifestyle, and biometrics data to locate the high-risk patients and assist preventive measures[20]. Nevertheless, generalizability has been a main concern because models trained on certain demographic groups can perform poorly when applied to other groups. The methodologies presented to enhance the scalability, cross-population, and privacy-sensitive training of collaborative models for predicting chronic diseases include transfer learning, domain adaptation, and federated learning.

3.4. Neurological Disorder Diagnosis

The IoT makes it easier to create AI-powered diagnostic instruments that are crucial for the early detection and therapy for neurological conditions such stroke, Alzheimer's, Parkinson's, and epilepsy. The DNNs are trained to manipulate MRI, CT, EEG, and cognitive assessment data to determine the subtle abnormalities in the brain's anatomical and functional characteristics. AI enhances early diagnosis, diseases progression prediction, and treatment planning on an individual basis. These methods improve diagnostic accuracy, reduce physician effort, and facilitate timely neurologic intervention.

3.5. Infectious Disease Detection

AI-based disease diagnostic systems significantly enhance infectious disease detection by predicting antimicrobial resistance and optimizing personalized antibiotic prescriptions using ML models trained on large-scale electronic health records. These systems improve treatment accuracy, reduce mismatched therapies, and support data-driven clinical decision-making[21]. Additionally, AI-powered chatbots utilizing NLP and hybrid learning approaches provide real-time symptom assessment, patient guidance, and outbreak-related information dissemination. Such applications strengthen epidemic preparedness, improve patient education, and streamline infectious disease management.

3.6. Genomic and Precision Medicine Applications

The use of AI in precision medicine and genomics can be used to conduct early risk stratification through the analysis of genetic risk factors (GRF), as well as combining genomic, clinical, and imaging data[22]. Deep learning models are also applicable in predicting the susceptibility to disease in the context of Alzheimer disease, cardiovascular diseases, type 2 diabetes, chronic kidney disease, and inherited cancers through the use of gene polymorphisms (e.g., APOE, BRCA1/2) and GWAS data. AI can be used to assist personalized screening, early intervention, and targeted treatment planning by integrating genetic biomarkers with electronic health records and images[23]. Such developments improve the accuracy of predictions and provide proactive and precision-oriented healthcare management.

Table II lists some of the most important AI-driven medical applications, explains their function, and cites pertinent research.

Table 2: AI Applications in Healthcare - Technologies, Benefits, and Use Cases

AI Application	Key Purpose	Technologies Involved	Key Benefits
Disease Diagnosis	Accurate diagnosis and early identification of conditions including cancer, heart disease, and neurological issues	NLP, CNNs, and deep learning	Enhances early intervention, lowers human error, and increases diagnostic accuracy.
Drug Discovery	Identification of molecular patterns, drug targets, and prediction of drug efficacy	Machine Learning, Reinforcement Learning, Computational Biology Models	Reduces time and cost in early-stage drug development; accelerates screening processes
Personalized Treatment	Customizes treatment plans using patient-specific genetic, lifestyle, and clinical data	Predictive Analytics, Genomic Data Integration, Machine Learning Models	Enhances treatment effectiveness, minimizes adverse drug reactions, supports precision medicine
Health Records Management	Automates Electronic Health Record (EHR) retrieval, analysis, and organization	NLP, Blockchain Technology, Data Mining	Improves administrative efficiency and ensures secure, tamper-proof access to patient data
Clinical Trial Optimization	Enhances patient recruitment, cohort selection, and clinical study design	Supervised Learning, Pattern Recognition, Predictive Modeling	Improves trial efficiency, reduces operational costs, and accelerates drug approval processes
Crowd-Sourced Health Data Analytics	Analyzes large-scale public health and population-level datasets	Big Data Analytics, Cloud Computing, Distributed AI Systems	Enables large-scale epidemiological research and strengthens public health monitoring
Outbreak Prediction and Monitoring	Predicts, tracks, and monitors infectious disease outbreaks using multi-source data	Machine Learning, Time-Series Forecasting, Predictive Models	Enhances epidemic preparedness, improves resource allocation, and supports early response strategies
Medical Imaging Analysis	Automated interpretation of diagnostic imaging, including MRIs, CT scans, and X-rays	Image processing methods, deep learning, and CNNs	Reduces diagnostic errors, enhances early diseases diagnosis, and supports radiologists

The analysis of non-synthetic data has been particularly useful in identifying diseases and drug development as well as individualized medicine. It is noteworthy that the possibility of significantly influencing the course of the health of the population and clinical results is significantly evident in its application in epidemic predictions and medical imaging diagnostics. Nonetheless, it is necessary to continue conducting studies on how to resolve concerns like algorithmic bias and data privacy to enable the appropriate development of AI's use in healthcare.

4. Challenges and Future Directions In AI-Based Disease Diagnostic Systems

The significant barriers to the large-scale adoption of the AI based disease diagnostic systems include the quality of the data, lack of standardization, interpretability, regulatory and infrastructure barriers. It also has in the future such prospects as AI-enabled robotics, oncology, digital pathology, integration of IoT and genomics, and AI-assisted clinical decision support to offer more specificity, accessibility, and efficiency in healthcare.

4.1. Challenges in AI-Based Disease Diagnostic Systems

There are still a number of diagnostic issues that need to be fixed before such technologies can be widely used. To achieve reliability, safety, and an ethical fit into the healthcare practice, it is necessary to address these barriers.

4.1.1. Lack of Standardization

The imaging protocols, pathology slide preparation, scanning parameters, and data annotation standards differ significantly and significantly influence the models' performance. Lack of standardized AI development and validation pipelines reduces reproducibility and cross-institutional generalizability.

4.1.2. Data Quality and Availability

Medical datasets are frequently dirty, incomplete, skewed, or inconsistently categorized. It is not because there is limited access to large, high-quality annotated datasets, which limits model robustness[24]. The limitation of data sharing among institutions also contributes to the impossibility of the creation of generalized AI systems.

4.1.3. Generalization and External Validation

Machines that were exclusively trained using datasets of a single centre or a homogeneous population often do not work well across different clinical environments. Inadequate multi-centric validation and real-world testing inhibit clinical translation.

4.1.4. Interpretability and Explainability

The decision-making mechanisms of many DL models are difficult to comprehend because they are black-box models. Poor explainability decreases the confidence of the clinician, and it presents difficulties to regulatory approval and clinical uptake.

4.1.5. Algorithmic Bias and Fairness

The AI systems might also reproduce the demographic and Socioeconomic biases in healthcare inequities that might arise from the training data[25][26]. The deployment of AI requires fairness and representative datasets to be ensured.

4.1.6. Regulatory, Ethical, and Privacy Concerns

AI-based diagnostic devices should align with stringent healthcare policies on patient privacy, data security, and informed consent. The ethical challenges include accountability, transparency, and the future of clinicians in the AI-assisted decision-making.

4.1.7. Interoperability and Data Standardization

A lack of standardized data among systems hinders the integration of healthcare systems. The use of the principles of Findable, Accessible, Interoperable, Reusable (FAIR) data and Common Data Elements (CDEs) is needed to promote the harmonization of data, its reproducibility, and collaborative research.

4.1.8. Hardware and Infrastructure Constraints

The implementation of AI also necessitates computing power, extensive storage capabilities, and reliable network connectivity[27]. The infrastructures are expensive, and the technical ability in a resource-constrained environment does not allow widespread use.

4.2. Future of AI In Disease Diagnostics

The advancement of new technology and the use of AI in disease diagnosis are expected to completely transform the medical field, interdisciplinary cooperation, and innovative practices. Outlined below are critical areas of improvement and steps to be taken in order to use AI to its full potential.

4.2.1. Advanced technologies in AI and their potential in disease diagnosis

- Robotics surgery: AI and ML-powered robot surgery is revolutionizing minimally invasive surgery. Robot bronchoscopy in case of lung cancer diagnosis and in case of endometriosis diagnoses have been verified to be effective in the Da Vinci system which is FDA-approved, with better accuracy and better patient outcomes. Building robotic technologies in the operating room that can make decisions in real time. Increasing the applicability of robotics driven by AI in under-served areas to enhance access to high-level surgical services.
- Oncology: The use of AI is transforming the care of cancer patients by providing them with a tailored approach to treatment. Timor microenvironments are being studied using ML models and predicting the response to treatment and subsequent biomarkers. Enhancing risk assessment and treatment plans with multi-omics data (genomes, proteomics, and metabolomics). Developing AI-powered tools to track the course of cancer and the efficacy of cancer treatment.
- Pathology: AI-powered digital pathology (DP) is using digital image analysis to replace conventional tissue biopsies. AI algorithms have been found to be better than human pathologists in detecting the metastasis of lymph nodes in breast cancer. Harmonizing DP processes to provide interoperability and consistency between healthcare systems. Combining immunohistology, histology, and molecular data with AI to create comprehensive diagnostic data.
- Skin Cancer: AI has demonstrated impressive levels of accuracy when classifying skin lesions, and DL models are at par with dermatologists. Creating multimodal AI platforms that utilize both imaging data, patient history, and genetic data to make the AI more accurate. Implementing AI-based diagnostic methods in the primary care environment to enhance the level of early diagnosis.

4.2.2. Synergy with other technologies: AI, genomics, IoT, and beyond

- AI and IoT: The healthcare industry may now employ predictive analytics and real-time observation to integrate AI and IoT. Patients' health outcomes can be accurately predicted using IoT sensors and AI algorithms. Creation of a safe IoT infrastructure to guarantee the privacy of data and avert hacker attacks. Scaling the IoT to resource-constrained and remote environments to provide decentralized healthcare.
- AI and genomics: AI is enhancing genomics through the identification of pathogenic variations and improved mapping of phenotype to genotype. The development of AI-based systems to develop personalized genomic medicine, where individuals receive unique treatment plans depending on their genetic composition. The integration of EHRs with genetic data provides comprehensive diagnostic insights.

4.2.3. AI's transformative impact on healthcare professionals and systems

AI is simplifying medical procedures by task automation including patient triage and scan interpretation. For instance, the American College of Cardiology has pioneered AI-driven chatbots have been used by the UK National Health Service for first diagnosis, while AI is being used to eliminate unneeded imaging. Creation of AI-based decision support systems that help with workload reduction and heightened accuracy of diagnostic results for clinicians. Adopting AI-based solutions in order to streamline resource distribution and minimize healthcare costs.

5. Literature Review

The literature states significant advances in the precision of clinical decision-making and diagnosis. However, there are a number of challenges still, especially in the area of strong validation, model interpretability, explain ability, and regulatory and ethical practices. A more detailed view of the recent research on disease diagnostic systems is presented in Table III, showing the main development of newer machine learning models to more advanced large language model (LLM)-based systems.

S. Zhou et al (2025) they conduct a thorough overview of the approaches to the application of the LLM in disease diagnosis. The paper goes through the literature that exists in several dimensions, such as types of disease and related clinical specialties, clinical data, the methods of LLM and assessment techniques as well. Moreover, they provide suggestions on how to use and assess LLMs to perform diagnostic work. Moreover, they evaluate the constraints of the existing studies and comment on the perspectives. This is as far as know the first extensive review of disease diagnosis based on LLM[28].

C. Dong, Y. Liu, J. Nie, X. Zhang, F. Yu, and Y. Zhou, (2025) based on a methodical search of databases, such as To summarize the current state of affairs, relevant research published between 2018 and 2025 were found using PubMed and Web of Science. It demonstrates the possibility of change, particularly in the field of diagnostic support. The creation of novel infectious diseases, pandemic control, and antibiotic resistance are just a few of the global issues that may be addressed with the help of AI technology. Utilizing its strong competencies in pattern recognition, data mining, and predictive analytics, AI supports early outbreak detection and intervention, improves the efficacy and accuracy of diagnostic processes, and offers real-time monitoring. Drawing from a survey of current literature, this narrative review methodically researches the use of AI in disease diagnostics[29].

A.-M. Alhejaily (2025) provides a thorough rundown of the developments made possible by AI in healthcare, explains how AI is now being utilized to enhance healthcare decision-making quality and efficacy as well as the healthcare system, and goes over a few specific medical uses of AI. Additionally, the potential future directions of healthcare systems enhanced by AI are explored, along with the obstacles and limitations that can prevent the application of AI in healthcare. The results of research show that interest in creating AI solutions for the healthcare sector is rising[30].

A. H. Rabie and A. I. Saleh (2024) provide a comprehensive approach to computer-aided disease diagnosis (CAD2) that is built on ML methods to assist physicians in developing better medical judgments. The three main sequential phases that comprise the proposed CAD2 are the phases of classification (CP), feature selection (FSP), and outlier rejection (ORP). The novel Outlier Rejection Technique (ORT), which consists of two successive steps named Fast Outlier Rejection (FOR) and Accurate Outlier Rejection (AOR), is used to reject outliers. Using the Hybrid Selection Technique (HST), the most informative characteristics are chosen using FSP. The two main stages of HST are the Quick Selection Stage (QS2) and the Precise Selection Stage (PS2), which use the Fisher score as a filter and Hybrid Bio-inspired Optimizations (HBO) as a wrapper. Lastly, CP uses the Ensemble Classification Technique (ECT) to actually diagnose the patient[31].

D. A. Ramalingam, D. A. Karunamurthy, D. T. Amalraj Victoire, and B. Pavithra (2023) provide a detailed examination of the many uses of AI in healthcare, including NLP, robotics, and ML. Additionally, the study examines potential uses of AI in healthcare, including medication discovery, disease prevention and prediction, and personalized therapy. Additionally, the study looks at the moral and legal conundrums that come with using AI in healthcare[32].

Y. Kumar, A. Koul, R. Singla, and M. F. Ijaz (2023) based on artificial intelligence techniques to diagnose numerous diseases, such as liver, skin, and kidney disease, diabetes, cancer, Alzheimer's, TB, stroke, cerebrovascular disease, and chronic heart disease. Conducted a comprehensive study that included the process of extracting and classifying features to make predictions, as well as the medical imaging dataset that was used. To locate papers on early disease prediction using AI techniques published up until October 2020, Psychology, the Web of Science, Scopus, Google Scholar, PubMed, and the Excerpta Medical Database are all subject to the Preferred Reporting Items for Systematic Reviews and Meta-Analyses standards[33].

Table 3: Literature Review on AI-Based Disease Diagnostic Systems

Authors (Year)	Study On	Methods	Key Findings	Applications	Limitations
S. Zhou et al. (2025)	Comprehensive examination of disease diagnostic techniques based on LLM	Systematic review of LLM applications; analysis across disease types, clinical specialties, datasets, LLM architectures, and evaluation	First comprehensive review focused exclusively on LLM-based diagnostic systems; provides structured recommendations for	LLM-driven diagnostic assistance, clinical text interpretation, multi-specialty disease	Limited clinical validation of LLMs; issues in explainability, hallucination risks, dataset bias, and lack of standardized

		metrics	applying and evaluating LLMs in clinical environment	diagnosis	evaluation frameworks
C. Dong et al. (2025)	AI applications in infectious disease diagnostics	Systematic and narrative review of studies (2018–2025) from PubMed & Web of Science; synthesis of AI techniques for infectious diseases	AI demonstrates transformative potential in outbreak detection, real-time monitoring, antimicrobial resistance prediction, and pandemic control	COVID-19 detection, tuberculosis diagnosis, outbreak forecasting, epidemiological surveillance	Limited generalizability across regions; dependency on high-quality real-time data; integration challenges with public health systems
A.-M. Alhejaily (2025)	Overview of AI advancements in healthcare systems	An extensive description of how AI is used in healthcare system optimization and decision-making	AI significantly enhances healthcare efficiency, decision accuracy, and patient outcomes; growing research interest in AI-based solutions	CDSS, predictive analytics, personalized treatment planning	Regulatory barriers, ethical concerns, privacy issues, infrastructure constraints in healthcare adoption
A. H. Rabie & A. I. Saleh (2024)	ML-Based Computer-Aided Disease Diagnosis (CAD2)	Three-phase ML framework: Outlier Rejection Phase (FOR + AOR), Feature Selection Phase (Fisher Score + Hybrid Bio-inspired Optimization), Ensemble Classification Technique	Improved diagnostic accuracy through hybrid feature selection and ensemble classification; robust outlier handling enhances reliability	General disease classification systems; ML-based structured clinical data diagnosis	Model complexity; computational overhead; limited discussion on real-time deployment and external validation
D. A. Ramalingam et al. (2023)	AI applications in healthcare (ML, NLP, robotics)	Comprehensive review of AI domains including ML, NLP, robotics; analysis of ethical and regulatory aspects	AI enables personalized medicine, disease prevention, predictive healthcare, and drug discovery; highlights future innovation areas	Disease prediction, robotic-assisted surgery, NLP-based medical documentation, preventive healthcare	Ethical dilemmas, regulatory ambiguity, absence of transparency, necessity for standardized frameworks
Y. Kumar et al. (2023)	AI-based diagnosis of multiple diseases	PRISMA-based systematic review; analysis of medical imaging datasets, feature extraction, and classification techniques	AI techniques (CNN, SVM, ANN, etc.) achieve high predictive performance across diseases as diabetes, heart disease, cancer, and Alzheimer's	Medical imaging-based diagnosis; early disease prediction; multi-disease classification	Dataset imbalance; overfitting issues; limited real-world clinical validation; heterogeneity in evaluation metrics

6. Conclusion and Future Work

Artificial intelligence disease diagnostics systems are transforming the healthcare industry by offering quicker, more precise, and tailored diagnosis features. Machine Learning performs best when working with organized clinical data, providing efficient classification and prediction and decision support, Deep learning makes it possible to analyze complicated imaging and signal-based data in a more accurate way. NLP can be used to promote automation of clinical data and patient data extraction. Applications of AI in medical imaging, chronic and cardiovascular diseases prediction, neurological disorders, infectious disease diagnoses, and Patient outcomes, treatment planning, and early diagnosis can all be improved by precision medicine.

Concerns around data quality, standardization, interpretability, bias, regulatory compliance, and infrastructure limitations remain significant despite these advancements. The barriers should be combated to ensure greater adoption. The uptake of cutting-edge technologies like IoT and genomics, and AI-assisted robotics will continue to improve healthcare delivery and streamline clinical processes and encourage proactive, precision-focused patient care.

In future studies, it is necessary to develop scalable, interoperable and explainable AI systems that incorporate multimodal data, including genomics, IoT, and clinical records. With the development of AI-based robotics, digital pathology, and personalized treatment systems, it will be possible to identify the disease at an early stage, provide support in decision-making in real-time, and offer equitable and precise healthcare to various groups of people with limited resources and opportunities.

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