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Modern Methods and Prospects for Using Artificial Intelligence in Disease Diagnostics: A Narrative Review

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Abstract

Aims: Assess AI methodologies in early disease detection, identify application areas like imaging and prognosis, determine key findings and clinical impacts, explore AI's prospects for accuracy and personalization, and analyze implementation challenges.

Methodology: This narrative review examines AI's role in medical diagnostics by analyzing peer-reviewed articles from 2019-2024 from PubMed. A comprehensive search strategy identified 338 publications, which were filtered for relevance, resulting in 10 key studies. Focus areas include AI techniques like machine learning and their applications and challenges in disease diagnosis.

Results: Results showed that in 2024, notable AI studies included a large experimental study with 27,558 samples, a comparative study with 1,653 samples, and a cross-sectional study with 20 samples. Previous years saw a retrospective cross-sectional study with 90,000 samples, an observational study with 1,052 samples, and a retrospective study with 219 samples. AI techniques featured EfficientNet-B2, CNNs, VGG-16, and ResNet variants, with transfer learning models like VER-Net and methods such as COVID-DSNet. Advancements highlighted EfficientNet-B2's 97.57% accuracy in malaria detection, VER-Net's superior lung cancer detection, and AI's effectiveness in diagnosing retinal diseases, heart conditions, diabetic nephropathy, and COVID-19. Challenges included computational demands and dataset needs, with recommendations for model optimization and clinical integration.

Scientific Novelty: This review is about the integration of advanced AI techniques in disease diagnostics, showing new algorithms and machine learning models that improve accuracy, speed, and personalized treatment strategies in medical practice.

Conclusion: This study has shown that there has been a significant progress in AI-based disease diagnostics, with examples of high performing models such as EfficientNet-B2 and VER-Net. Despite challenges like computational requirement and interpretability of the model, AI has the potential to revolutionize diagnosis.

Keywords: deep learning, transfer learning, machine learning, model optimization.

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Introduction

AI has transformed different industries with healthcare being a standout among the most encouraging areas[1]. In health care, disease diagnostics is very important and will benefit greatly from AI's data analysis capacity, pattern recognition as well as predictive modeling. Diagnostic accuracy may be heightened and the rate of diagnostic errors can be minimized if AI is integrated into diagnostics [2, 3]. Accurate diagnosis is important in disease management and ensuring patients' wellness. General errors by human beings reduce the accuracy of diagnoses, particularly in general clinical practice and rural areas, due to their complicated and cognitively demanding nature when interpreting biomedical information [4]. Artificial intelligence approaches such as CNNs, knowledge graphs, have been shown to be powerful and highly encouraging for diagnosis and management of diseases. The use of AI in diagnostics assists doctors to enhance diagnostic accuracy levels as well as efficiency hence providing emergent digitalized healthcare services [5, 6].

AI technologies especially ML and DL have demonstrated excellent efficiency in interpreting complicated medical information. Therefore, they are used in diverse diagnostic tools such as image analysis and predictive analytics for pathology and genomics among others [7, 8]. Moreover, AI algorithms are more effective than human experts in detecting abnormalities in medical images. Also, through analysing large datasets using AI it is possible to identify hidden patterns and connections that could be missed by conventional diagnostic approaches [9, 10]. AI's potential benefits in the field of diagnostics go far beyond improving accuracy and efficiency. AI-based tools can help identify diseases at earlier stages, contributing to prompt intervention and treatment. This is especially true for ailments like cancer that require early detection for successful therapies. Besides, AI also contributes in personalized medicine by adopting diagnostic as well as therapeutic strategies tailored to specific patients depending on their genetic, environmental and life-style characters [11, 12].

AI, which is guided by algorithms and runs on computers, has been programmed to learn from data itself and give intelligent forecasts as well as real-time resolutions using ML, ANNs, robotic process automation and data mining [13,14]. The WHO has admitted that technology greatly helps in uniting long-term care with the daily lives of patients. Technology becomes very important when achieving universal health coverage for people of different age groups through efficient service delivery and cost-effective integration. For instance, the use of AI and robotics in technological development is gaining popularity due to its support in healthcare research as well as practice [15]. The integration of AI in healthcare has the potential to revolutionize patient care by providing accurate diagnoses and personalized treatment plans. Furthermore, advancements in AI technology can lead to improved health outcomes and enhanced efficiency in healthcare systems.

However, there are also some problems with using AI in disease detection. This includes the concern of inadequate data privacy, lack of transparent algorithms and vital need for rigorous validation studies to ensure ethical and safe use of AI in clinics [16, 17]. To add on, healthcare professionals must be educated and trained to use these tools effectively as it demands a change in thinking from conventional teaching methods in the health workforce towards adoption of AI technologies [18,19]. The task can be intimidating for healthcare institutions to create AI systems that are ethical and safe, which may involve large expenses in the management of data, control of algorithm, and an analysis of societal and environmental effects of the AI [20, 21]. The process of embedding responsible AI demands interdisciplinary and cross-sector collaborations among data scientists, health care providers and policy makers with mutual interests in creating a sustainable AI ecosystem that is beneficial to all stakeholders and society [22, 23]. The integration of AI into healthcare is not merely a technical challenge but also an educational and cultural one. Ensuring that AI tools are used responsibly and effectively requires comprehensive training and a shift in mindset among healthcare professionals.

Transparency has often been mentioned as a key problem to be solved if AI is to be accepted, regulated and deployed in healthcare. In other words, it deals with whether AI algorithms and models can be explained or verified [24, 25]. AI has transformed significantly since Christopher Strachey developed the first AI program in 1951. At that point, AI was still young, and it formed an area of academic interest for many researchers. In 1956 John McCarthy organized a meeting at Dartmouth College where he invented the expression "artificial intelligence." This occurrence marked the start of modern AI. From 1960s to 1970s, AI research concentrated on expert systems and rule-based reasoning. However, this approach suffered from inadequate data and computing capacities [26].

The present research is aimed at presenting a general perspective on AI in disease diagnostics, focusing on current trends, major achievements and obstacles. In view of the interaction between technological innovation and clinical practice, we intend to reveal how promising the prospects for AI in changing disease diagnostics are, which will result in good medical care.

Research Problem

Incorporation of artificial intelligence in the field of disease diagnostics is a situation that offers immense opportunities and has therefore necessitated detailed examination. One of the main problems under investigation is how well AI systems such as ML, CNNs, NLP, Predictive Analytics, and DL are able to correctly identify diseases. Although they have demonstrated potential, it is necessary to assess if these technics surpass old diagnostic methods through scientific methodology.

Moreover, another major concern is the existence of differences in the types of data and sample sizes used across different researches. Different kinds of clinical data, medical images, patient records, electronic health records, and genomic data have their respective pros and cons. It is therefore necessary to comprehend how these types of data influence AI diagnostics' dependability and performance so that a standardized procedure can be established. Still, this raises serious research questions regarding how study outcomes are influenced by changes in sample size as well as the applicability of results. Also, there is a need to scrutinize such research issues that examine in detail why and under what conditions it is statistically meaningful to relate study outcomes to population parameters through samples involving small numbers.

Also, there are several impediments to the practical AI implementation in clinical settings. For instance, data privacy issues, high computational costs, biases in NLP models, integration challenges with existing healthcare systems and complexity in interpreting genetic data act as barriers to the wide scale adoption of AI. These obstacles must be surmounted so as to ease the transition from idea stage to a fully functional AI program for medical use.

Research Focus

The study will concentrate on a thorough examination of current AI techniques employed in diagnosis of diseases so as to determine their effectiveness and find out ways of enhancing them. A critical review of various AI modes like Machine Learning, CNNs, NLP, Predictive Analytics and Deep Learning will be carried out to compare their diagnostic accuracy, efficiency and applicability across different clinical settings. This can only be done through a detailed analysis of studies involving different data types like clinical data, medical images, EHR, patient records and genomic data thus understanding the impact of these variables in predicting AI performance.

Additionally, this research will also look into clinical applications of AI and how these technologies improve early diagnosis of illness, better medical imaging, make textual analysis easy for clinicians, help in prognosis of diseases as well enable personal medication. By combining major conclusions drawn from other studies, the paper will show the clinical effect of AI like- enhanced diagnostic accuracy; decision support system, treatment planning and personalized medicine based on genetic profiling.

In addition, the research will also consider future of AI in diagnosing diseases and problems that arise. This includes looking at potential improvements in the accuracy and speed of diagnosis, creating more advanced imaging methods, improving patient records analysis, better predictions of risk and managing it having a basis on genetic make-up as well as personalizing treatment plans grounded on genes. Furthermore, the study will list down possible solutions to present-day issues such as; privacy concerns regarding information sharing online, processing costs of computer intensive AI models to mitigate biasing factors in decision making processes, compatibility between new AI systems and legacy IT infrastructure within firms' context and finally how data analysis tools can be used you translate genomic data into clinical action so that they can be adopted by relevant health care providers.

The objectives of this research are to give a broad outline of the current status of AI in disease diagnosis with focus on these areas, to detect areas where information is lacking and provide recommendations for future studies that aim at improving the field.

Research Aim

1. Evaluate diverse ways of applying AI in the early detection and diagnosis of illnesses.
2. Point out some areas where AI has been used such as medical imaging, clinical text analysis, disease prognosis and personalized medicine regarding early disease detection.
3. What are some key facts found in research on AI application to diseases' diagnosis?

4. How does AI improve diagnostic accuracy, imaging techniques and management while developing treatment plans for individual patients?

5. Consider the difficulties encountered when implementing AI into practice in detecting or diagnosing diseases.

Research Questions

1. What are the different approaches used in AI for early detection and diagnosis of diseases?

2. Where has AI been deployed, including early disease detection, medical imaging, clinical text analysis, disease prognosis or personalized medicine?

3. How have AI applications impacted on the findings and clinical outcomes in diagnostic processes?

4. In what ways can AI contribute to better management and personalized treatment plans, as well as improve imaging techniques and enhance diagnostic accuracy?

5. What are some of the obstacles faced by health experts regarding implementation of AI in disease detection and diagnostics?

Research Methodology

General Background

Modern disease diagnosis methods are recently using artificial intelligence to improve precision and productivity. Complex medical data, like imaging, and genetic information is analyzed by AI technologies that include deep learning and machine learning to determine the patterns of diseases and their outcomes. These advancements have immense opportunities for early detection, personalized therapy, and enhanced results for patients. The transformative character of AI in healthcare is based on its ability to process vast datasets as well as integrate numerous diagnostic modalities. There is current research being done on optimizing AI algorithms, overcoming issues such as data integrity & interpretability, and ensuring fairness in accessing AI-driven test innovations.

Study Design

A narrative review of peer-reviewed original articles published from 2019 to 2024.

Search Strategy

The literature search was performed utilizing a combination of keywords to explore the use of artificial intelligence in diagnosis of disease using keywords from PubMed database ("Artificial Intelligence" OR "AI") AND ("Disease Diagnosis" OR "Disease Detection" OR "Medical Diagnostics") AND ("Machine Learning" OR "Deep Learning" OR "Neural Networks" OR "Support Vector Machines" OR "Classification Trees" OR "Predictive Analytics") AND ("Current Trends" OR "Future Prospects" OR "Applications" OR "Challenges").

Study Selection

Conducting a comprehensive literature search in PubMed constituted the search strategy and at first, 338 publications relating to the research topic were initially identified. This was followed by screening of records with respect to certain criteria; among these, 21 were excluded for not being within the years 2019 to 2024, 94 because they lacked free full-text access, 125 due to not involving human subjects and others irrelevant numbering 63. Once those filters were applied on the records, only thirty-five (35) articles were perceived as eligible for further assessment thus ten (10) highly relevant studies have been included in the final review as depicted in Figure 1.

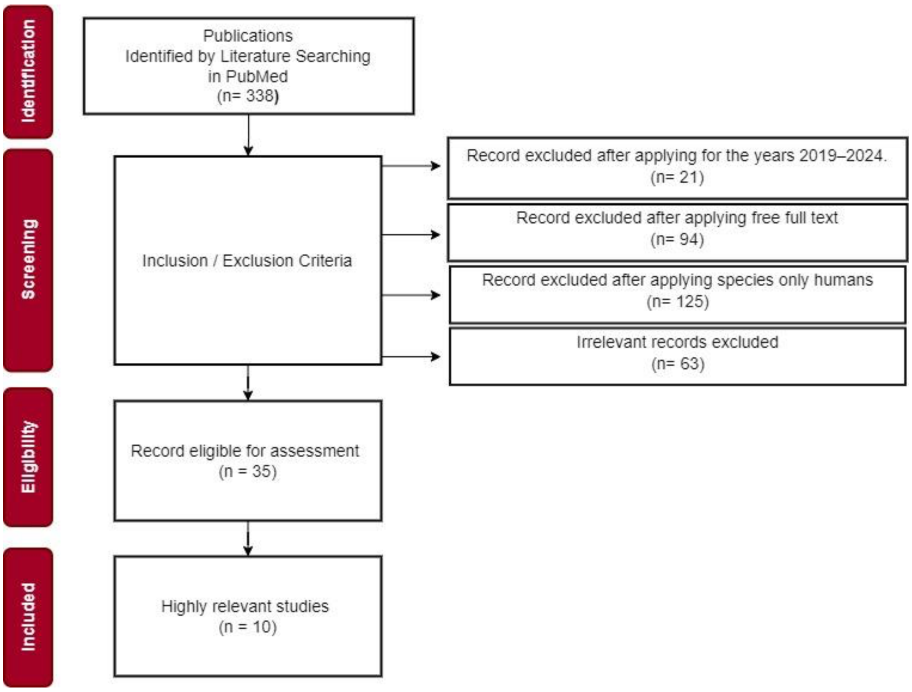

Figure 1. **PRISMA flow diagram**

Source: authors' development

Data Analysis

The data analysis for the study on modern AI methods in disease diagnostics involves assessing the accuracy, efficiency and performance of different AI models and techniques. This includes comparing deep learning, transfer learning, ensemble methods, and specific architectures. Challenges such as computational needs, dataset limitations, and integration issues are also examined.

Research Results

The utilization of artificial intelligence (AI) in disease diagnostics has seen significant advancements and diverse research methodologies. Studies spanning from experimental to observational designs demonstrate the broad application and potential of AI to enhance diagnostic accuracy and efficiency in the medical field. Table 1 summarizes the finding of current study on AI techniques in medical diagnostics. Research [27] achieved 97.57% accuracy with EfficientNet-B2. Another study [28] found VER-Net superior in accuracy and sensitivity. The integration of VGG19, EfficientNetB0, and ResNet101 was highlighted in research [29]. Additionally, research [30] showed deep learning's high accuracy in retinal disease detection. Combining sensor-based auscultation and ECG with machine learning, study [31] demonstrated high diagnostic efficiency. Machine learning was also used for early diabetic nephropathy diagnosis in research [32]. A multi-input deep learning model improved classification accuracy, as noted in study [33]. Further studies [34] and [35] demonstrated high accuracy with deep learning and ensemble methods. High accuracy with ensemble methods and stochastic models was achieved in research [36]. In conclusion, these studies collectively demonstrate the evolving capabilities of AI and machine learning in enhancing diagnostic accuracy across various medical fields. The integration of advanced techniques and models is consistently improving performance, suggesting a significant shift toward more reliable and efficient diagnostic tools

Table 1. **Study characteristics**

Author's / Year	Study Design	Sample Size	AI Technique	Key Findings
Mujahid et al. 2024 [27]	Experimental	27558	Deep Learning	EfficientNet-B2 achieved 97.57% accuracy
Saha et al. 2024 [28]	Comparative study	1653	Transfer Learning-Based Deep Learning Techniques	VER-Net outperforms other models in accuracy, sensitivity, and specificity
Isangula et al. 2024 [29]	Nonexperimental cross-sectional	20	Transfer Learning-Based Deep Learning Techniques	VER-Net model integrates VGG19, EfficientNetB0, and ResNet101, showing superior performance

Rodríguez-Cobo et al. 2023 [30]	Retrospective cross-sectional study	90000	Deep Learning	High accuracy in detecting retinal diseases, superior to traditional methods
Shiraga et al. 2023 [31]	Observational study	1052	Machine Learning Models	High diagnostic efficiency with sensor-based auscultation and ECG combined with machine learning
Su et al. 2023 [32]	Retrospective study	219	Machine Learning Models	Machine learning models with ultrasound imaging and biochemical data effectively diagnose early DN
Reis et al. 2022 [33]	Experimental	2,357	Deep Learning	COVID-DSNet outperformed other models in accuracy and classification
Ruengchaijatuporn et al. 2022 [34]	Cross-sectional study	918	Deep Learning	Multi-input model with soft labels and self-attention improves classification accuracy
Daniel et al. 2022 [35]	Prospective, observational cohort study	1066	Deep Learning	High segmentation and diagnostic accuracy, outperforming traditional methods
Arora et al. 2021 [36]	Experimental study	5840	Ensemble Methods and Stochastic Models	High accuracy and AUC for X-ray images, better than CT images

Source: authors' development

Table 2 summarizes the finding comparing various AI techniques and models for different diagnostic tasks. Deep learning models achieved 97.57% accuracy for malaria detection [27]. Transfer learning-based deep learning techniques demonstrated high accuracy with VER-Net, achieving 91% and 95.2%, respectively [28,29]. Ensemble methods combining CNNs reached 92.5% accuracy [30]. Machine learning models, including stacking models, were used for heart conditions with varying accuracy [31]. KNN, SVM, and LR were compared, with KNN achieving 91% [32]. High accuracy for COVID-19 detection was reported using deep learning techniques [33]. VGG16 achieved 81% accuracy [34]. UNet++ achieved 94.75% accuracy for segmentation [35], and ensemble methods with stochastic models reached 91% overall accuracy for X-ray images [36]. The results of this study show that deep learning and transfer learning models are effective in different diagnostic tasks, often achieving accuracy rates above 90%. Ensemble methods and specific architectures such as UNet++ and VGG16 also exhibit strong performance, indicating the adaptability and potential of these techniques in medical imaging and diagnosis.

Table 2. The performance of various AI models and techniques

Author's	Algorithms/Models	AI Technique	Accuracy of the AI model
Mujahid et al. 2024 [27]	EfficientNet-B2, CNN, VGG-16, DenseNet121, DenseNet169, DenseNet201, Inception V3, ResNet50, EfficientNet-B1, EfficientNet-B7, MobileNet, MobileNetV2	Deep Learning	97.57% accurate in detecting Malaria
Saha et al. 2024 [28]	VER-Net (integrating VGG19, EfficientNetB0, ResNet101); other models for comparison.	Transfer Learning-Based Deep Learning Techniques	VER-Net scored 91% accuracy
Isangula et al. 2024 [29]	VER-Net, integrating	Transfer Learning-Based Deep Learning Techniques	95.2% accuracy
Rodríguez-Cobo et al. 2023 [30]	Convolutional Neural Networks (CNNs) Transfer Learning with VGG16, ResNet50, and DenseNet121 Ensemble Methods combining different CNN architectures	Deep Learning	92.5% accuracy
Shiraga et al. 2023 [31]	Neural Networks: 10-layer Convolutional Neural Networks (CNN) for heart sound and ECG data. Stacking Models: Random Forest (RF) and XGBoost (XGB) for combining outputs of individual CNN models.	Machine Learning Models	Severe Aortic Stenosis 93%, Severe Mitral Regurgitation 80% and Left Ventricular Dysfunction 75%
Su et al. 2023 [32]	K-Nearest Neighbor (KNN) Support Vector Machine (SVM) Logistic Regression (LR)	Machine Learning Models	KNN 91%, SVM 84% and LR 84%
Reis et al. 2022 [33]	COVID-DSNet, COVID-DSNet + FCC, COVID-DSNet + LSTM, InceptionResNetV2, InceptionV3, MobileNet, ResNet-101, DenseNet-169, NASNetMobile, EfficientNetB0.	Deep Learning	CT dataset: COVID-DSNet achieved 97.60% CXR dataset: COVID-DSNet achieved 88.34%

Ruengchaijatuporn et al. 2022 [34]	VGG16, multi-input VGG16, Conv-Att with soft labels and self-attention.	Deep Learning	81% accuracy
Daniel et al. 2022 [35]	UNet++ for segmentation; comparison with U-Net.	Deep Learning	94.75%
Arora et al. 2021 [36]	DenseNet, GoogleNet, VAE, Support Vector Machine (SVM), Random Forest (RF), XGBoost (XGB), Logistic Regression (LR).	Ensemble Methods and Stochastic Models	91% overall for X-ray images

Source: authors' development

Table 3 highlight the significant advancements in disease diagnostics through artificial intelligence (AI). The EfficientNet-B2 model achieved a 97.57% accuracy in malaria detection from red blood cell images, offering timely diagnoses in areas with limited skilled microscopists [27]. The VER-Net model, integrating VGG19, EfficientNetB0, and ResNet101, outperforms other models in lung cancer detection, enhancing diagnostic accuracy and treatment planning [28, 29]. AI has been shown to be superior in diagnosing retinal diseases compared to traditional methods, leading to earlier interventions and better outcomes [30]. High diagnostic efficiency for valvular heart diseases was achieved using a machine learning approach combining sensor-based auscultation and ECG [31]. Machine learning models with ultrasound and biochemical data have proven effective for early diabetic nephropathy diagnosis [32]. The COVID-DSNet model excels in accuracy and classification for pneumonia and COVID-19 detection [33]. A multi-input model with self-attention improved early detection of Mild Cognitive Impairment [34]. High accuracy was achieved in diagnosing eosinophilic esophagitis from biopsy slide images, and X-ray images provided a faster, scalable diagnostic tool for COVID-19 and pneumonia compared to CT scans [35, 36].

Table 3. Applications and Outcomes

Author's	Application Area	Clinical Impact
Mujahid et al. 2024 [27]	Malaria detection in red blood cell images	Precise, prompt diagnosis in areas lacking skilled microscopists
Saha et al. 2024 [28]	Lung cancer detection using CT scan images	Enhanced diagnostic accuracy for lung cancer
Isangula et al. 2024 [29]	Medical Imaging, specifically in the detection and diagnosis of lung cancer	Improved early diagnosis and treatment planning for lung cancer
Rodríguez-Cobo et al. 2023 [30]	Diagnosis and management of retinal diseases	Earlier intervention and improved patient outcomes
Shiraga et al. 2023 [31]	Diagnosis of valvular heart diseases and left ventricular dysfunction	Enhanced early diagnosis and treatment of valvular diseases
Su et al. 2023 [32]	Early diagnosis of diabetic nephropathy in T2DM patients	Facilitates early intervention and improved patient outcomes
Reis et al. 2022 [33]	Diagnosing pneumonia and COVID-19	Improved early detection and patient outcomes for COVID-19 and pneumonia
Ruengchaijatuporn et al. 2022 [34]	Early detection of Mild Cognitive Impairment (MCI)	Accurate early detection aiding timely prevention and intervention
Daniel et al. 2022 [35]	Eosinophilic esophagitis diagnosis using esophageal biopsy slide images	Efficient EoE diagnosis, reducing diagnostic time and improving accuracy
Arora et al. 2021 [36]	Diagnosis of COVID-19 and pneumonia using chest X-ray and CT scan images	Fast, scalable diagnostic tool for medical facilities

Source: authors' development

Table 4 provides a comprehensive overview of the prospects and challenges associated with modern AI methods in disease diagnostics. It highlights key studies, such as those emphasizing large-scale clinical implementation but facing high computational resource needs, suggesting future directions like dataset enlargement and parallel computing [27]. Another study focuses on dataset expansion and detection systems but encounters high computational demands, recommending model combinations and transfer learning [28]. A different study discusses model improvement through transfer learning while facing data limitations and generalization issues, advocating for dataset enlargement and model experimentation [29]. Integration into clinical workflows is addressed in another study, which struggles with 'black box' issues and dataset biases, suggesting diverse datasets and interpretable models [30]. Multimodal AI applications and their performance decline in clinical settings are explored, with recommendations for model optimization and clinical integration [31]. Another study outlines that there is a revolution in AI diagnostics, which points out data quality and model generalizability as concerns with suggestions for future research on dataset enlargement and clinical validation [32]. Improvement in medical imaging is mentioned, which has to grapple with the challenges of small datasets and noisy data while recommendations are made on increasing model depth and data augmentation [33]. Broader clinical applications are emphasized in a different study, grappling with 'black box' nature and generalizability, recommending improved model interpretability and deep learning advancements [34]. Extending AI applications to various conditions is another focus, with struggles in managing large image data, proposing dataset expansion and model refinement [35]. Finally, expansion to other imaging domains is highlighted, facing computational complexity and data management issues, with future directions aimed at dataset expansion and clinical integration [36].

Table 4. Prospects and Challenges

Author's	Prospects	Challenges	Future Directions
Mujahid et al. 2024 [27]	Large-scale clinical implementation	High computational resource needs, such as GPU clusters and extensive training times (e.g., weeks to months)	Enlarge dataset using data augmentation, implement parallel computing with distributed systems
Saha et al. 2024 [28]	Dataset expansion and detection systems	High computational needs, dataset size limits (e.g., requiring petabytes of storage and terabytes of RAM)	Model combinations with ensemble methods, transfer learning using pre-trained models
Isangula et al. 2024 [29]	Model improvement with transfer learning	Data limitation, model generalization (requiring high-quality labeled datasets and advanced pre-trained models)	Dataset enlargement via synthetic data generation, model experimentation with different architectures
Rodríguez-Cobo et al. 2023 [30]	Integration into clinical workflows	'Black box' issue, dataset biases (necessitating explainable AI methods and diverse training datasets)	Create diverse dataset with varied demographics, develop interpretable models using techniques like SHAP and LIME
Shiraga et al. 2023 [31]	Multimodal AI applications	Performance decline in clinical settings (requiring specialized hardware like TPUs and robust cross-validation)	Optimize models for specific clinical tasks, integrate models into clinical workflows with continuous feedback loops
Su et al. 2023 [32]	AI revolution in diagnostics	Data quality, model generalizability (demanding high-resolution imaging and extensive data augmentation)	Expand datasets through partnerships with healthcare providers, validate models with clinical trials
Reis et al. 2022 [33]	Development in medical imaging	Limited dataset, noisy data (requiring advanced denoising algorithms and large annotated datasets)	Increase model depth using deeper neural networks, apply data augmentation techniques like GANs for synthetic data generation
Ruengchaijatuporn et al. 2022 [34]	Broader clinical application	'Black box' nature, generalizability (necessitating interpretable models and comprehensive validation studies)	Improve model interpretability with explainable AI methods, enhance deep learning models with continuous learning
Daniel et al. 2022 [35]	Application to other conditions	Expanding EfficientNet-B2's dataset with synthetic images improves diversity and balances precision and recall for accurate eosinophil counting	Expand dataset using federated learning, refine models with transfer learning from similar datasets
Arora et al. 2021 [36]	Expansion to other image domains	Computational complexity, data management (needing distributed computing frameworks and scalable data pipelines)	Enlarge dataset with cloud-based storage solutions, integrate models into clinical workflows with edge computing

Source: authors' development

Discussion

The current study summarizes various included studies on AI in disease diagnostics. These include a large-scale experimental study, a comparative study, and a small nonexperimental cross-sectional study. Additionally, there are retrospective and observational studies, as well as a prospective observational cohort study. Previous research also features experimental studies. These studies highlight diverse approaches and extensive research in applying AI to improve disease diagnostics.

The current study reviews AI techniques in disease diagnostics, detailing advancements in deep learning, transfer learning, and machine learning models. Deep learning methods include EfficientNet-B2, CNN, VGG-16, DenseNet, Inception V3, ResNet50, and MobileNet. Transfer learning models encompass COVID-DSNet, InceptionResNetV2, NASNetMobile, UNet++, and VER-Net. Machine learning models feature neural networks, Random Forest, XGBoost, K-Nearest Neighbor, Support Vector Machine, and Logistic Regression. Ensemble and stochastic methods discussed include DenseNet, GoogleNet, VAE, and Random Forest. Recent studies from 2019 to 2024 illustrate these models' effectiveness and advancements in diagnosing various diseases. Similarly, another study emphasizes the role of deep learning in automating the evaluation of medical images, leading to more accurate diagnoses and reduced human error in image interpretation and illustrates how deep learning algorithms can analyze complex data patterns, making them suitable for tasks like cancer detection and other critical disease diagnoses [37]. The integration of these AI techniques as we move forward is expected to revolutionize disease diagnostics, thus improving precision and efficiency in medical practice. The ongoing evolution of these technologies highlights the potential for transformative improvements in healthcare outcomes.

COVID-DSNet and InceptionResNetV2 are two transfer learning models that have become popular during the COVID-19 pandemic, highlighting the significance of AI in disease diagnosis and treatment. The study also found out that these models use pre-trained networks to improve performance on certain tasks with limited data. Additionally, another research evaluates how efficient machine learning-based predictive approaches including transfer learning methods are when it comes to cardiovascular ailment detection and detecting their suitability for clinical environments [38–40]. The above developments show how AI can change medical diagnostics. As we improve these technologies, the use of AI in clinical practice will help to improve accuracy and efficiency in disease detection and management.

This study also emphasizes the importance of machine learning in many diseases, indicating that these models are often more accurate and reliable than traditional diagnostic tools and also emphasizes the need for choosing appropriate algorithms based on the individual disease context. According to previous research on this subject, Machine Learning techniques such as Random Forest, XGBoost, K-Nearest Neighbor (KNN), Support Vector Machine (SVM), Logistic Regression (LR) have remained critical in diagnosis of diseases and speed up the process of disease management [41,42]. This discussion stresses that the use of these sophisticated algorithms can greatly improve diagnostic accuracy and efficiency, thus leading to better patient outcomes and call for further research and improvement of these instruments in order to maximize their potential in clinical settings.

Additionally, the current research underscored that the AI methods integration into disease diagnostics is rapidly evolving, with significant indications of viability depicted by deep learning, transfer learning and machine learning models in improving accuracy and efficiency of diagnosis. The above technologies are expected to play important role in healthcare as a result of technological advancements in this field, which will eventually lead to enhancement of patients' condition through accurate and timely discovery of different diseases. Disease diagnosis using artificial intelligence has made significant strides in enhancing accuracy and efficiency across numerous medical fields. Similarly, there was another study which found out that AI models could effectively detect various cardiovascular diseases demonstrating the potential of machine learning to improve diagnostic outcomes within resource-challenged contexts. This concurs with EfficientNet-B2 results implying that AI might help bridge healthcare access disparities through availability of consistent diagnostic tools for underserved locations [43, 44]. As mentioned in the discussion, the integration of AI into diagnostics not only simplifies the process but also has the potential to democratize healthcare access by ensuring that everyone has access to high-quality diagnostics.

In current study, the VER-Net model that consists of VGG19, EfficientNetB0 and ResNet101 has shown an improvement in lung cancer detection. In addition, other researches pointed to the increased use of convolutional neural networks (CNNs) for cancer diagnostics specifically breast and lung cancers where AI models such as VER-NET can increase diagnostic accuracy by analyzing intricate images facilitating earlier interventions and more accurate treatments [45, 46]. As mentioned earlier, the integration of advanced CNN architectures such as VER-Net significantly enhances the efficiency of AI in medical imaging. This development is a significant step towards more precise and timely cancer diagnosis.

Current study also highlighted that AI's superiority over traditional methods in diagnosing retinal diseases has been corroborated by various studies included. Similarly, other recent studies explored the application of deep learning algorithms in ophthalmology, showing that AI could significantly reduce the time required for diagnosis and improve the accuracy of detecting conditions like diabetic retinopathy and this advancement supports the assertion that AI leads to earlier interventions, which is critical for preserving vision and improving patient outcomes [47,48]. The results of this study show that AI can change ophthalmology. It can improve the accuracy of diagnosis and make it easier for doctors to diagnose patients. As AI develops, it could be used in everyday medical practice to help identify retinal diseases earlier and treat them more effectively, which would result in improved patient care and outcomes.

Current study also showed that the COVID-DSNet model's success in detecting pneumonia and COVID-19 is part of a broader trend where AI has been pivotal during the pandemic. Similarly, other studies have also shown that AI-enhanced diagnostic tools, such as those analyzing X-ray images, can provide rapid and accurate assessments compared to traditional CT scans and this capability was proved crucial during the COVID-19 outbreak, as timely diagnosis could significantly impact treatment outcomes [49, 50]. The development of this technology shows that AI is increasingly being used to manage diseases effectively and can change the way we diagnose diseases in future health emergencies. Moreover, the use of AI technologies can improve healthcare efficiency and patient outcomes by enabling quicker and more accurate diagnoses.

Despite the fact that disease diagnostics have improved significantly in recent years through application of artificial intelligence (AI) with numerous studies pointing out its possible contribution to improving accuracy, efficiency and finally patient outcomes, several challenges and limitations need to be overcome before AI can be fully utilized in healthcare. One big challenge is having high quality and diverse training datasets for AI models. In the same vein, research calls for standardization as well as appropriate datasets creation to ensure successful integration of AI into disease diagnostics [51, 52]. Another study on the other hand underscores the importance of data quality issues like unlabeled data lack or biasness in pathology and radiology to enhance performance of AI systems [53, 54]. Another challenge identified in the study is interpretability and explainability of AI models, often referred to as “black box” problem. This has been reiterated by another research which emphasizes on the demand for more transparent and interpretable AI models in diagnosing breast cancer to gain trust from medical practitioners and patients through employing saliency mapping and adversarial training techniques to improve model interpretability [55, 56]. In conclusion, it is important to overcome these difficulties in order to achieve the maximum benefits of AI in healthcare. To improve diagnostic accuracy and patient outcomes, it is important that researchers, practitioners and policymakers work together to create strong, transparent and fair AI systems.

These challenges are also addressed by contemporary study by discussing some ethical and legal issues that should be considered when implementing AI in healthcare. This argument is supported by other studies that argue for strong ethical frameworks and regulatory policies to guide the responsible and equitable application of AI in monitoring inflammatory bowel disease and highlight the need for resolving problems such as data privacy, security, and algorithmic bias [57, 58]. Yet, AI can do much more than this given its potential in disease diagnostics. Different studies have identified a different approach to predicting diabetes through deep learning using clinical data transformed into image data that shows how versatile AI technology is in dealing with intricate diagnostic issues [59, 60]. To ensure that AI is effectively integrated into healthcare, it is important to strike a balance between technological innovation and strong ethical guidelines that guarantee patient safety and fairness. As AI advances, its capacity to handle intricate diagnostic challenges will increase, which could change the way diseases are predicted and managed.

To conclude, there are various issues that should be addressed to ensure that AI can be successfully implemented in the medical field despite its potential benefits. Among them are ethical and regulatory concerns, data quality problems, interpretability issues, as well as necessity of ongoing innovation and collaboration between AI scientists and medics. Once these obstacles are tackled head-on, we will begin to exploit AI’s comprehensive aptitude for disease diagnosis thereby advancing the welfare of patients.

Conclusions

In this study, there is an overall description of the present status and prospects of AI in future in disease diagnostics. The different designs used in various studies as well as the sample sizes and sources of data being published show how far we have gone and how much more we can do to incorporate AI in medical diagnostics.

The methodologies employed for AI are on display here with examples on deep learning, transfer learning and machine learning techniques. Models such as EfficientNet-B2, VER-Net among others convolutional neural network (CNN) and ensemble methods illustrated high performances across different diagnostic tasks. For example, the malaria detection model’s accuracy at 97.57% is a clear pointer to its potentiality towards improving diagnosis accuracy in cases where resources are limited like resource-limited settings. Another illustration can be drawn from the case of lung cancer detection using the VER-Net model which indicates that integration of several neural networks leads to better diagnostic accuracy.

AI has outperformed traditional methods in several diseases' diagnoses ranging from retinal diseases or valvular heart diseases to early recognition of conditions like diabetic nephropathy or eosinophilic esophagitis.AI can rapidly diagnose diseases using X-rays, biopsy images and other type imaging data compared to conventional means with greater efficiency.AI has been found superior in various aspects of diagnosis from ocular pathologies to valvular heart disorders; identification of early stages for certain complications like diabetic nephropathy or eosinophilic esophagitis etc., based upon photo data including X-rays, biopsies etc., demonstrates its capacity to develop timely scalable diagnostic solutions that sometimes supersede classical techniques both concerning precision and speed.

Despite these promising advancements, there are some challenges associated with the modern AI methods discussed here. They include but not limited to; huge computational resource requirements, data limitations, black box nature of AI models and clinical adoption hurdles. These challenges will be tackled by increasing dataset sizes, refining the model and improving its interpretability. Datasets should be expanded in future research, models optimized and clinical integration improved to bring AI closer to reality in diagnostics. Taken together, the growing body of knowledge shows that AI can revolutionize early-stage detection as well as treatment planning for disease diagnosis. The development and improvement of AI models may lead to more precise, efficient, and available diagnostic tools that could make a difference in patients' lives by enhancing outcomes and advancing medical science. Although significant advances have been made towards using AI for disease diagnostics, there is still much work needed to overcome current obstacles so that these techniques are widely adopted in clinical practice. In addition, these findings support the belief that AI will play a major role in diagnostics with the potential for significant improvements in diagnostic accuracy and patient care.

Implications

The results highlight the potential of artificial intelligence in disease diagnosis, thus enhancing accuracy, efficiency and accessibility across a range of medical conditions. The integration of advanced AI models like EfficientNet-B2 and VER-Net into clinical practice promises significant improvements in early disease detection and personalized treatment planning. Nevertheless, issues such as high computational costs, limited data availability and black-box AI models should be dealt with. However, if robust dataset expansion is achieved, along with model performance optimization and interpretation enhancement; success in integrating AI will have been accomplished. It is recommended that further studies will address these technologies while developing means of smoothly incorporating artificial intelligence into clinical workflows. This way AI can change forever diagnosis in medicine to enable a future where early detection and personalized care are the rule rather than the exception leading to better patient outcomes and advancement in medical science.

Suggestions for Future Research

To fully exploit the potential of artificial intelligence (AI) in diagnosing diseases, future research should focus on several key areas. First, it is important to expand and diversify datasets. Large representative datasets with diverse demographic and geographic information will enhance the robustness and generalizability of AI models. Collaborative efforts between institutions and countries can facilitate data sharing and the creation of comprehensive datasets. Secondly, model interpretability needs to be improved. The "black box" nature of many AI models is a major obstacle to clinical adoption. Developing explainable AI (XAI) techniques that produce transparent and interpretable results will help clinicians understand and trust AI-driven decisions. Research should aim at developing models that not only perform well but also provide insights into their decision-making processes. Thirdly, computational efficiency must be optimized for AI tools to be used in resource-constrained settings. Research should explore lightweight AI models and algorithms that require less computational power without compromising accuracy. This includes investigating edge computing and federated learning to process data locally and reduce reliance on centralized, high-resource servers. The main focus should be on integration into clinical workflows. Developing AI models that seamlessly integrate with existing electronic health record (EHR) systems as well as other healthcare infrastructure will facilitate smooth adoption. Pilot studies in clinical environments can help identify practical challenges and refine AI tools for real-world use. Additionally, ethical considerations and bias mitigation must be addressed. Research should ensure that AI models do not perpetuate or exacerbate existing healthcare disparities. This involves scrutinizing training data for biases and developing algorithms that promote fairness and equity in diagnostics. Finally, interdisciplinary collaboration is key. Engaging experts from AI, medicine, ethics, and healthcare policy will ensure that research is comprehensive enough to address the multifaceted challenges associated with integrating AI into disease diagnostics. Through these efforts, AI can become a transformative tool in healthcare, improving early detection, diagnosis, and personalized treatment across a wide range of diseases.

Declarations

Author Contributions

Conceptualization, Rayisa Yuriy and Olha Tatarina; methodology, Rayisa Yuriy; software, Rayisa Yuriy; validation, Rayisa Yuriy, Olha Tatarina and Valery Kaminskyy; formal analysis, Rayisa Yuriy; investigation, Rayisa Yuriy; resources, Rayisa Yuriy; data curation, Rayisa Yuriy; writing—original draft preparation, Rayisa Yuriy; writing—review and editing, Rayisa Yuriy; visualization, Rayisa Yuriy; supervision, Rayisa Yuriy; project administration, Rayisa Yuriy and Olha Tatarina.

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