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The Use of Artificial Intelligence in Disease Diagnosis: A Systematic Literature Review

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ABSTRACT

This paper discusses the crucial role of Artificial Intelligence in improving disease diagnosis and the use of medical data in the era of big data. Through the Systematic Literature Review (SLR) approach, we review recent developments in the application of Machine Learning (ML) to disease diagnosis, evaluate the ML techniques applied, and highlight their impact on healthcare. BP-CapsNet uses the Convolutional Capsule Network (CapsNet) to diagnose cancer with advantages in overcoming invariance to image transformation. The Stacking Classifier achieves 92% accuracy in detecting heart defects with the advantage of combining weak learners. CraftNet, a combination of deep learning and handmade features, demonstrates strong recognition capabilities in cardiovascular disease. ML in infectious diseases demonstrates the ability to process big data, focusing on bacterial, viral, and tuberculosis infections. In heart disease diagnosis, ML, especially with CNN and DNN, detects disease at an early stage, despite the challenges of data imbalance. The ensemble algorithm for heart disease prediction demonstrates the superiority of categorical medical features, with SVM and AdaBoost as suitable methods. A new CNN for wide QRS complex tachycardia provides accurate results. In vestibular disease, five ML algorithms provide satisfactory results, with SVM as the best. These findings detail the development of ML in disease diagnosis, highlighting future challenges and opportunities in its use.

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1. INTRODUCTION

Medical data collected in hospitals can be utilized by clinical decision support systems to assist doctors in diagnosing diseases more accurately, as well as to monitor the treatment process in real-time remotely [1]. The utilization of medical data must go through a data analysis process, such as Artificial Intelligence, Machine Learning, Data Mining, etc [2]. In addition to providing opportunities for the development of self-care services, the more medical and diagnostic data generated, the more data in various formats will pose a challenge, known as Big Data [3].

In the era of big data, various data-driven methods that are mostly based on Deep Learning (DL), have been applied in the field of diagnosis. Disease diagnosis has become one of the most important aspects of healthcare, with technological advancements being key in improving precision and efficiency as well as minimizing errors in the diagnosis process. Sometimes, the biggest challenge in healthcare is the late detection of diseases which can ultimately result in fatal consequences for individuals. Diseases detected early can often be prevented or stopped. In this context, the importance of Machine Learning-based disease prediction [4][5] is becoming increasingly significant as it can identify common diseases at an early stage, enabling earlier and more effective intervention [6].

The use of Machine Learning (ML) in disease diagnosis has been an important milestone in fundamental changes in the field of healthcare. In recent decades, ML has overhauled the paradigm of disease diagnosis, opening a window to revolutionary advances that offer more precise, efficient, and transformative approaches[7][8].

The importance of using Machine Learning in disease diagnosis has been a major concern in various studies. As suggested in previous studies classification methods using [9][10] Machine Learning algorithms offer a potential solution for early diagnosis of diseases. The study highlights that algorithms such as K-nearest, Decision Tree, Random Forest, Support Vector Machine (SVM), and Naïve Bayes have shown success in accurately predicting disease, with the potential to minimize diagnostic errors [11][12].

ML is not only an additional tool in the field of healthcare but is also an important foundation for the latest developments in disease diagnosis. Its ability to accurately process complex medical data has revolutionized the way we understand, diagnose, and manage health. The implementation of ML in disease diagnostics provides an undeniable edge for healthcare providers [13][14], suitable for complex applications [15]. With artificial intelligence, ML enables careful analysis of complex medical data, resulting in disease prediction and identification with an unprecedented degree of accuracy [16].

However, constraints in processing, storing, and analyzing historical data involving the continuous flow of data from healthcare have been a major challenge. This highlights the new complexity in medical data management, which goes beyond the storage capacity of traditional databases. Medical diagnosis, as one solution in this context, has a crucial role to play in addressing this challenge. It includes an important process in translating observational evidence into accurate disease diagnoses [17].

In a practical context, the existence of ML has brought about fundamental changes in the healthcare system as a whole. By processing large and diverse volumes of data, ML enables early detection, more precise diagnosis, and more focused, personalized care [18][19]that is safe and effective[20]. This not only improves clinical outcomes for patients but also optimizes the efficiency of the healthcare system. However, it was said in previous research [21] that the integration of machine learning methods with traditional methods will have a major impact on health services. ML is focused on making predictions as accurate as possible, whereas traditional statistical models are aimed at inferring relationships between variables.

The main purpose of this Systematic Literature Review (SLR) paper is to describe recent developments in the use of ML for disease diagnosis. This paper will comprehensively present various ML techniques applied in disease diagnosis, evaluating the effectiveness and accuracy of these technologies in specific disease cases. In addition, focus will also be given to the impact of ML in the context of healthcare as a whole, including its impact on costs, system efficiency, and benefits to the wider population [22].

Through this in-depth analysis, the SLR paper aims to provide a deeper understanding of how ML has shaped and continues to enrich the disease diagnosis landscape. It is hoped this exposure will be a useful first step towards understanding the extent to which ML has revolutionized disease diagnosis and its potential for further change in the future, as well as providing valuable guidance for future research and healthcare improvement [23].

2. METHOD

The methodology used is a systematic review, which is a methodology for finding relevant studies, selecting and investigating those studies, analyzing data, and summarizing findings to reach appropriate conclusions. A systematic literature review (SLR) is used to transparently and reproducibly summarize scientific evidence to answer a specific research question. This approach seeks to combine all published evidence on a topic and evaluate the quality of that evidence [24][25].

Deep literature searches are conducted in several databases, including IEEE Xplore at 10%, Google Scholar at 10%, PubMed at 20%, and Science Direct at 60%. The search was conducted using a series of relevant keywords, such as "Disease Diagnosis," "Machine Learning," "Artificial Intelligence," "Deep Learning," and "ML approach." This approach is designed to ensure that relevant and up-to-date research is identified for further investigation.

3. PREVIOUS WORK

The application of Machine Learning for the diagnosis and prognosis of various diseases with various methods has been described in several research papers, here is a summary of papers related to this:

Table 1. Results of Previous Research Review

Title	Author	Method	Disease	Result	Strength	Weakness
BP-CapsNet: An image-based Deep Learning method for medical diagnosis	[26]	Convolutional Capsule Network (CapsNet)	Cancer	BP-CapsNet can provide better results in diagnosing medical conditions from medical images, such as the detection of cancer or other diseases, compared to other Machine Learning methods. BP-CapsNet is also able to address some of the problems faced by CNN, such as invariance to image transformation and the ability to learn better representations of objects in images.	High computational complexity on BP-CapsNet, which can require large computational resources to train and run these models. In addition, the interpretation of the prediction results generated by CapsNet models may be more difficult compared to other traditional Machine Learning models.	CapsNet's ability to learn better representations of objects in images, invariance to image transformations, and ability to overcome some of the disadvantages of traditional Machine Learning methods.
A stacking classifier model for detecting heart irregularities and predicting Cardiovascular Disease	[27]	Stacking Classifier	Cardiovascular Diseases (CVDs), or heart diseases	Using 10 classification types, including agency, probabilistic, and ensemble, the model achieves 92% accuracy. MLP (Multi-Layer Perceptron) model, the proposed model achieves high accuracy and good sensitivity in identifying patients with heart disease.	The advantage of the model lies in the incorporation of weak learners through stacking techniques, resulting in stronger predictive results than traditional classifications.	The limitations of the datasets used, both in terms of the amount of data and attribute variation, may affect the generalization of the model for diseases other than the heart. High reliance on 10 different classifiers, as well as a focus on pre-processing and normalization

						techniques, can also be an obstacle and pose a risk of overfitting.
Machine Learning for clinical decision support in infectious diseases: a narrative review of current applications	[28]	Supervised learning, unsupervised learning, and reinforcement learning	Various infectious diseases, including bacterial infections, viral infections, tuberculosis, and other infections	The results showed that of the 60 ML-CDSS found, most focused on bacterial infections (62%), viral infections (17%), and tuberculosis (15%). Further, 33% of them were aimed at diagnosis of infection, 30% for prediction, early detection, or stratification of sepsis, and 22% for prediction of response to treatment. However, of those 60 ML-CDSS, only 57 reported performance measures such as sensitivity or specificity, and there has not been sufficient evidence regarding their use and impact in real-life clinical settings.	His ability to process and analyze large volumes of data, as well as his ability to learn from data and interpret unknown situations	Limitations in real-life clinical evaluations, reliance on data from high-income countries, and lack of information regarding interactions between physicians and ML-CDSS in real-life clinical settings.
CraftNet: A deep learning ensemble to diagnose cardiovascular diseases	[29]	CraftNet, an approach that combines deep learning and "handcraft" features	Cardiovascular Disease (CVD)	CraftNet, the proposed method, was tested on the MIT-BIH public dataset. The experimental results showed strong performance with sensitivity	CraftNet integrates handmade features and deep learning, addressing the problem of data imbalance. The use of decision-directed cyclic graphic	The issue of data imbalance in ECG signals affects deep learning performance, although it has been partially addressed by handmade features.

				reaching 88.16%, 85.37%, 94.53%, and 88.92% for the four categories tested. The average sensitive accuracy also increased from 86.82% to 89.25%. CraftNet is considered to have strong recognition capabilities and is less affected by data imbalance.	topology (DDAG) gives good results.	
Machine Learning-based heart disease diagnosis: A systematic literature review	[30]	Metode Machine Learning seperti Naive Bayes, Decision Tree, Support Vector Machines (SVM), dan Deep Learning (DL) such as Convolutional Neural Network (CNN) dan Deep Neural Network (DNN)	Cardiovascular diseases such as coronary heart disease, cerebrovascular disease, peripheral artery disease, rheumatic heart disease, and congenital heart disease	This systematic study of the literature concluded that ML, especially when applied to electrocardiogram (ECG) and patient data, can help detect heart disease at an early stage. However, the main challenge faced is data imbalance in heart disease data sets, which can hinder traditional ML performance.	ML helps improve data-driven decision-making in heart disease diagnosis. Improved performance of ML models, especially with the use of DLs such as CNNs and DNNs. ML's flexibility and ability to leverage patient and ECG data.	The problem of data imbalance in heart disease data sets is a serious challenge. ML models and DL-based solutions can lack a proper explanation of model behavior during final predictions. The performance of ML algorithms can tend to be biased towards majority classes, especially on unbalanced datasets. The challenge of interpretation and explanation of ML algorithms, especially in Deep Learning-based models.
Impact of categorical and numerical	[31]	Gradient Boost, Extreme	Cardiovascular disease (CVD)	The results showed that categorical	The use of various ML methods	There are challenges in collecting

features in ensemble Machine Learning frameworks for heart disease prediction		Gradient Boost, AdaBoost, CatBoost, logistic regression, decision tree, random forest, artificial neural network, and support vector machine (SVM)		medical features were more effective in CVD prediction compared to numerical features or a combination of both. Ensemble classifiers using SVM and AdaBoost proved suitable for heart disease prediction. The performance of this method was evaluated using various performance metrics and resulted in the finding that categorical features predominate in prediction accuracy.	provides flexibility and the ability to explore different aspects of medical data. Ensemble learning with SVM and AdaBoost improves prediction accuracy.	limited and inconsistent medical data, affecting ML algorithm training. Method performance is highly dependent on the availability of high-quality data, and some patient features may be missing or unavailable. Some ML algorithms have high computational complexity, which can be a bottleneck in scenarios with limited resources.
A novel convolutional neural network structure for differential diagnosis of wide QRS complex tachycardia	[32]	Convolutional Neural Network (CNN)	Wide QRS complex tachycardia (WCT)	The proposed CNN model achieves a detection accuracy of 87.5% for VT and 91.7% for SVT. Using 5-fold cross-validation and running the algorithm for five independent runs, the sensitivity, specificity, positive predictive value, negative predictive value, and F1 score of the CNN model were approximately 88.50%.	The CNN model achieves a high degree of accuracy in classifying VT and SVT in patients with WCT. CNN models have the potential to be used in real-time settings, assisting clinicians in interpretation, and decision-making.	Limitations of data in the representation of clinical case variation, reliance on training data that may not cover a wide range of clinical conditions, and the potential for errors in human diagnoses used as ground truth
Application of Machine	[33]	Random Forest	Peripheral vestibular	The five tested Machine	All five Machine Learning	Optimal results were obtained

Learning in the diagnosis of vestibular disease		(RF), Adaboost (AB), Gradient Boosting (GB), Support Vector Machine (SVM), Logistic Regression (LR)	disease (PV) and non-PV disease	Learning algorithms gave satisfactory results. The accuracy of the algorithm ranges from 76 to 79%, with the Support Vector Machine (SVM) classification having the highest accuracy. In cases where the prediction results of all five models were consistent, the accuracy of PV diagnosis results was improved to 83%, while for non-PV diagnosis results increased to 85%.	algorithms provided satisfactory results, demonstrating the potential use of Machine Learning in supporting the diagnosis of vestibular disease. Relatively high accuracy, especially with the use of Support Vector Machine (SVM). Research provides evidence that Machine Learning can help in the classification of vestibular diseases based on the results of balance function tests.	with a limited number of patients (1009 patients). As a suggestion for future research, the authors suggest increasing the number of patients to improve diagnosis accuracy. Optimization of classification methods was also identified as a potential area of development to achieve higher diagnostic accuracy.
Artificial intelligence applications in restorative dentistry: A systematic review	[34]	Artificial intelligence , machine learning	restorative dentistry	The accuracy of AI models in diagnosing dental caries ranging from 76% to 88.3%, with sensitivity ranging from 73% to 90%, and specificity ranging from 61.5% to 93%. The accuracy of caries prediction ranging from 83.6% to 97.1%. The accuracy of vertical tooth fracture diagnosis ranging from 88.3% to 95.7%. The accuracy of detecting the tooth	AI models show the potential to be a powerful tool in assisting with the diagnosis of dental caries and vertical tooth fracture, detecting the tooth preparation margin, and predicting restoration failure.	Despite the potential shown by AI models, the research states that the application of AI in dentistry is still in development. This presents opportunities for future research to: Assess the clinical performance of AI models more comprehensively. Explore ways to enhance the accuracy and precision of AI models.

				preparation finishing line ranging from 90.6% to 97.4%.		Investigate better integration of patient data for AI model training. Understand and address ethical concerns or constraints associated with the use of AI in dentistry.
Meta-Analysis of the Performance of AI-Driven ECG Interpretation in the Diagnosis of Valvular Heart Diseases	[35]	Convolutional Neural Network (CNN)	Valvular Heart Diseases (VHD)	AI-driven ECG shows high accuracy in VHD screening with the following outcomes: Pooled accuracy of 81% (95% CI 73 to 89, I^2 = 92%). Sensitivity of 83% (95% CI 77 to 88, I^2 = 86%). Specificity of 72% (95% CI 68 to 75, I^2 = 52%). Positive predictive value (PPV) of 13% (95% CI 7 to 19, I^2 = 90%). Negative predictive value (NPV) of 99% (95% CI 97 to 99, I^2 = 50%).	AI methods in ECG interpretation provide high accuracy, sensitivity, and specificity, indicating significant potential for early detection of Valvular Heart Diseases (VHD).	Despite the advantages of AI-driven ECG, the low positive predictive value (PPV) suggests the need for a combined approach with clinical judgment, especially in primary care settings. Therefore, opportunities for future research may include: Development of improved AI models to enhance PPV. Integration of additional data, such as clinical information, to improve diagnostic accuracy. Further research on ECG recording variability and methods to address such issues. Ethical and clinical acceptance research on AI in healthcare settings.

Based on a review of several papers in the table, we can identify common patterns that reflect progress and challenges in applying Machine Learning in medical diagnosis. Some papers use deep learning approaches such as Convolutional Neural Network (CNN) and Capsule Network. Capsule Network shows advantages in understanding the representation of objects in medical images, while CNN is widely used and achieves high accuracy, especially in diagnosing cardiovascular diseases.

Ensemble learning approaches, such as stacking classifiers and combining handmade features with deep learning, are emerging as effective strategies for improving model accuracy. Although they provide more robust predictive results, some papers note high computational complexity, which requires large computational resources. Data imbalance in heart disease datasets is a common challenge. Traditional Machine Learning models can be biased towards majority classes, and reliance on many different classifiers in classifier stacking can increase the risk of overfitting.

The limitations of datasets, both in terms of the amount of data and the variety of attributes, are a major concern. Most studies note the importance of high-quality data and consistency in medical datasets. The limited number of patients may affect the generalization of models for diseases other than those studied. Several papers highlight difficulties in interpreting model prediction results, especially for deep learning models. The limitations of the interpretation and explanation of Machine Learning algorithms, especially in deep learning models, are aspects that need more attention.

In general, research in this area shows positive progress in improving the accuracy of medical diagnosis using Machine Learning. However, challenges such as data imbalances, dataset limitations, and model interpretation remain key focuses in further development. The application of ensemble learning and the emphasis on high-quality data can be a promising solution to overcome some of these obstacles.

4. MODEL CREATION STAGE

Data Collection

In the stage of collecting data for disease diagnosis using artificial intelligence (AI), the identification of medical resources such as electronic medical records, medical image results, and relevant clinical data is essential. This process provides the data base needed to train and test AI models that will be used in supporting disease diagnosis.

Data Processing

Data that has been collected through the collection stage undergoes a series of processing steps. Data cleaning, normalization, and feature extraction are performed to ensure that the data used in AI model creation is well processed, free of anomalies, and ready for use in the training process.

Model Selection

In the context of disease diagnosis, AI model selection is becoming a key aspect. This stage involves selecting the AI model that best fits the needs of the Clinical Decision Support System (CDSS) for disease diagnosis. Consideration involves the type of model, such as classification or regression model, that is most effective in analyzing specific medical data for diagnostic purposes.

Model Training

After model selection, the AI model is trained using a processed dataset. This training process involves teaching models to recognize patterns and important information in medical data. The goal is to ensure that the model can make decisions accurately and reliably in the context of disease diagnosis.

5. RESULTS AND DISCUSSION

5.1 Utilization of Medical Data in Clinical Decision Support Systems

Previous research suggests information can be a tremendous resource for the healthcare industry and the benefits associated with data. Medical analytics across various healthcare organizations has helped create considerable attention to Healthcare informatics and analytics (HCI&A). Big data shows great potential to support a wide range of medical functions and healthcare services such as clinical decision support, disease surveillance, and public health management [36][37].

The industry is faced with the need to combine medical knowledge or clinical rules with advancements in Machine Learning that offer high predictive accuracy, along with the increasing application of Artificial Intelligence technology [38]. The utilization of medical data in Clinical Decision Support Systems using Machine Learning (ML) can be described in Figure 1 [39].

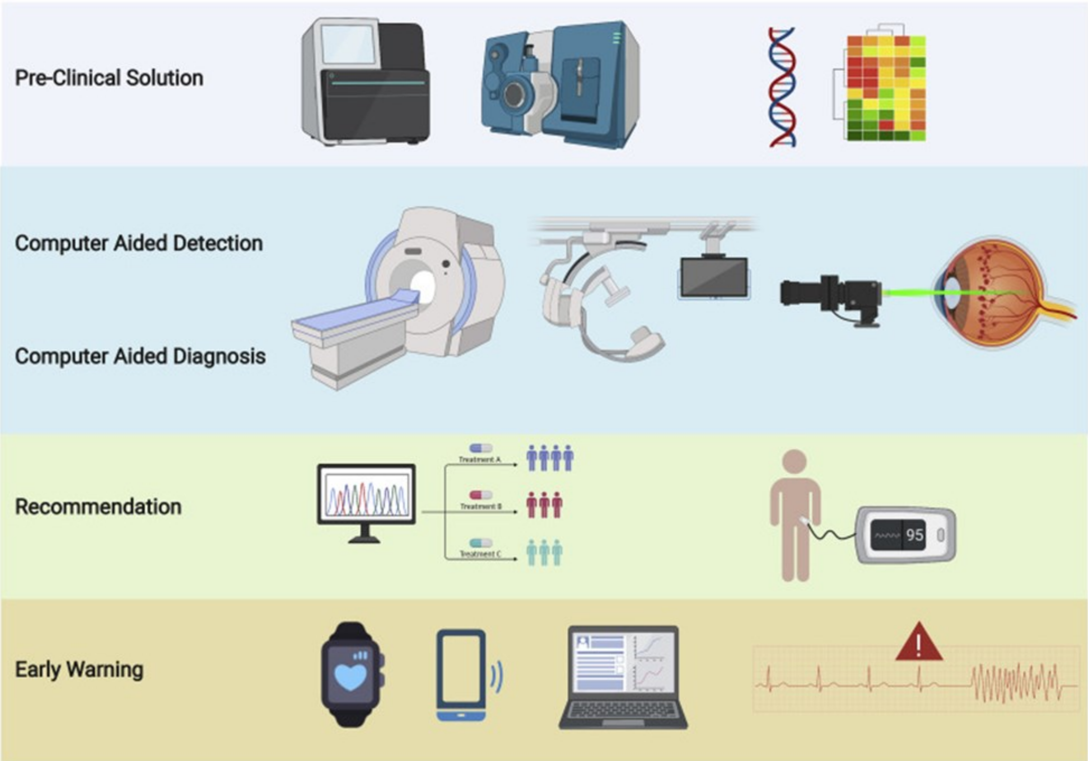

Figure 1 Graphical representation of ML specification in clinical decision making

1. Pre-Clinical Solutions (Clinical Introduction)
   In the pre-clinical phase, ML is utilized to analyze genomic and metabolome data through DNA and metabolome sequencing devices. The goal is to find biomarkers that can provide deep insight into a patient's condition and support better clinical decision-making.
2. Computer-Aided Detection (CAD) for Medical Image Detection
   ML is applied to medical image acquisition techniques such as magnetic resonance, X-rays, and retinal photography. CAD systems use ML to detect and analyze medical images, assisting doctors in diagnosing diseases more accurately.
3. Precision Medicine Drug Recommendation
   ML is used in developing a drug recommendation system tailored to the individual characteristics of the patient. This includes analysis of clinical and genetic data to provide drug recommendations that are more precise and tailored to each patient's unique needs.
4. Early Warning through Smart Wearable and Electronic Health Record Monitoring
   ML is leveraged in real-time health monitoring through smart wearables and electronic health records. ML algorithms can continuously identify patterns and anomalies in health data, provide early warning of potential health risks, and support rapid clinical decision making.

In artificial intelligence (AI)-based clinical decision support systems (CDSS), medical data is used to improve diagnosis accuracy and monitor the remote treatment process. CDSS integrates various medical records, literature, and clinical research data to evaluate drug efficacy, product availability, adverse reactions, patient financial status, and types of medical insurance. By leveraging big data and Machine Learning, CDSS can provide personalized individualized advice to clinicians, helping them optimize treatment plans. For example, the Watson for Oncology system provides standard treatment methods for specific cases in a short period of time based on the case data entered. CDSS such as the Chinese Society of Clinical Oncology-Artificial Intelligence (CSCO AI) has also been used to provide services at a national level in breast cancer treatment, helping patients achieve personalized treatment based on drug approval and treatment guidelines. By harnessing medical data holistically, CDSS supports more effective clinical decision-making, improves diagnosis accuracy, and provides guidance based on the latest scientific evidence, especially in the context of telemedicine.[40]

Artificial Intelligence (AI) refers to the use of computational methods that allow machines to perform tasks such as perception, reasoning, learning, and decision-making. It has been argued in previous papers that disease diagnosis is a critical aspect of treatment planning and patient care success, but is often hampered by human error because the task of understanding medical information is complex and cognitively challenging. The application of AI in disease diagnosis aims to overcome this obstacle and has shown great potential in improving diagnostic accuracy. The use of [41]Artificial Intelligence (AI) in disease diagnosis provides various advantages, especially through Machine Learning methods such as Supervised, Unsupervised, and Deep Learning that have been integrated into the diagnostic process. Through these methods, medical data can be processed to produce predictions, identify patterns, and understand data associations tailored to patient needs and the treatment process [42].

Machine Learning is a computer algorithm that learns from data to make future predictions without explicit programs. In the era of big data, Machine Learning is popular because it can analyze big data. The integration of Machine Learning in clinical imaging promises task automation, improved diagnosis, and the potential discovery of new biomarkers [43]. Machine Learning is seen as a more dynamic approach to problem-solving with computers compared to traditional computer programming [44].

Specifically in a previous paper, it was explained that [45] Machine Learning (ML) has a significant role in the transformation of various industries, including the health sector. ML is used to process and analyze large and diverse health data, resulting in accurate predictions and intelligent solutions for a wide range of health tasks.

Some applications of ML in health contexts

1. Diagnosis. ML is used to analyze electronic medical records (EHRs), perform clinical feature extraction, and support the diagnosis process. The use of ML in medical image analysis helps in disease detection, organ classification, image segmentation, and medical image reconstruction.
2. Prognosis. ML can be used to predict disease progression, health risks, and treatment outcomes based on patient health data.
3. Therapy. ML helps in devising an effective and personalized treatment plan according to the individual characteristics of the patient.
4. Clinical Workflow. ML improves the efficiency of clinical workflows by providing advice to radiologists and physicians, assisting in decision-making, and automating some routine tasks

While ML has brought advancements, there are concerns about the security and robustness of ML models, especially in the context of health. Adversarial attacks and data privacy concerns are challenges that need to be addressed to ensure secure and reliable ML deployments in healthcare. That way, ML has the potential to transform the way diagnosis and healthcare are performed, providing faster, more accurate, and personalized solutions.

5.2 The Role of Machine Learning in Diagnosis

The main challenge in medical services today is the delay in diagnosis of diseases that can have a negative impact on treatment outcomes. This problem is not just limited to specific cases, but rather includes a complex range of diseases. Research from on the Clinical Impact of Late Diagnosis of Hirschsprung's Disease in Newborns, states that late diagnosis of Hirschsprung disease (HD) in newborns will have an impact on increasing the risk of serious complications such as Hirschsprung-associated enterocolitis (HAEC) and failure to thrive. It is mentioned in this study that early diagnosis is very important and necessary to provide appropriate treatment to prevent adverse complications [46].

Other studies specific to the diagnosis of primary immunodeficiencies (PIDs) stated similarly, that delays in diagnosis will cause delays in the administration of appropriate treatment, and more severe can increase the risk of death as the duration of delay in diagnosis increases. The paper also agrees that with early diagnosis in patients, patients can be given appropriate treatment faster and have an impact on improving the quality of life of patients [47].

One of the causes of this delay can be attributed to the complexity of medical data which takes a long time to be processed and analyzed manually. The diagnosis process, which is one example of medical decision-making, involves heterogeneous and extensive medical data, so it often takes excessive time or even leads to an inability to handle such complex data. The right clinical decisions require a deep understanding of patient data, and manual processes are often inefficient in the face of this complexity. Therefore, an innovative approach is needed to speed up and improve the accuracy of the diagnosis process [48].

The solution to overcome this challenge lies in the utilization of Machine Learning (ML) technology. ML has been used in healthcare and medicine for a variety of disease diagnoses, leading to the development of decision support tools and other applications, such as drug prescription [49]. Machine Learning, with its ability to process and analyze medical data at scale, enables manual identification of patterns and relationships that are difficult to see. The use of ML can reduce the time required to diagnose diseases and increase the accuracy of diagnosis, which in turn contributes to the improvement of overall medical services.

By applying this approach widely, leveraging medical data through ML can not only address delays in diagnosis but also open up new opportunities for a deep understanding of various health conditions.

5.3. The Contribution of Machine Learning in Healthcare

Big data shows great potential to support a wide range of medical and healthcare functions such as clinical decision support, disease surveillance, and public health management. Rapid advances in patient Electronic Health Records (EHRs), and integration of social, behavioral, and omics data with mHealth, eHealth, Smart Health, and ICT-based telehealth devices have led to the development of new healthcare frameworks to support precision medicine and personalized patient care.

In the healthcare sector, the implications of big data are predictive analytics techniques and Machine Learning platforms for the provision of sustainable solutions such as the implementation of personalized treatment and medical care plans. The use of omics and clinical big data helps build predictive models for the understanding of biological processes and the design of effective drugs. Effective analysis of health data from big data sources helps pharmaceutical companies measure drug outcomes through smaller, shorter trials. The utilization of medical data through distributed technology can make a positive contribution to healthcare. The application of ML helps in the identification and treatment of various diseases, including urgent health challenges such as cancer or epidemic situations such as COVID-19. It can provide evidence-based support for medical interventions [50].

The role of Machine Learning in disease diagnosis, particularly through medical image processing, is in the spotlight. The utilization of this technology provides the ability to understand and analyze complex medical data with high accuracy, assisting physicians in making more informed and efficient decisions. Nonetheless, some challenges need to be addressed, including data security, privacy, and imperfections of the models used. The evaluation of methods related to various aspects, such as advantages, challenges, databases, implementation, privacy, and security issues, becomes key in optimizing the use of this technology in the world of health [51][52].

6. CONCLUSION

In the context of disease diagnosis, Machine Learning (ML) has proven a significant role in improving accuracy, efficiency, and early detection. The utilization of ML in Clinical Decision Support Systems (CDSS) opens the door to more precise diagnosis and remote monitoring, optimizing treatment plans and providing evidence-based support. Despite positive progress, challenges such as data imbalances, dataset limitations, and model interpretation continue to be the focus of development. Merging ML methods with traditional approaches offers potential solutions, but a thorough evaluation is needed to address aspects such as data security and privacy.

Suggestions for future research include further exploration into the integration of Machine Learning with current technologies such as blockchain to improve the security and privacy of medical data. A deeper understanding of how ML can address the challenges of data imbalance and dataset limitations is also a critical aspect to investigate. Further research can be directed at developing interpretable ML models to increase trust and adoption by health practitioners. In addition, further exploration of the impact of ML in the treatment of specific diseases and performance comparisons between various ML algorithms will provide richer insights into technology development in healthcare.

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