

**AI IN DISEASE DIAGNOSIS IN MACHINE LEARNING AND DEEP LEARNING AND THEIR APPLICATION AND CHALLENGES**

**Mr. M.I. Yasar Ihdisham<sup>1\*</sup>, Mr. R. Jagadesh<sup>1</sup>, Mr. R. Dhanush<sup>2</sup>, Mr. M. Premkumar<sup>2</sup>,  
Mr. M. Praveenkumar<sup>3</sup>, Mr. C. Jothimanivannan<sup>3</sup>**

<sup>1</sup>Student, SS Institute of Pharmacy, Sankari, Salem -637301.

<sup>2</sup>Assistant Professor, Department of Pharmacology, SS Institute of Pharmacy, Sankari, Salem  
– 637301.

<sup>3</sup>Professor & Principal SS Institute of Pharmacy, Sankari, Salem-6373901.

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**\*Corresponding Author****Mr. M. I. Yasar Ihdisham**

Student, SS Institute of Pharmacy,  
Sankari, Salem -637301.



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**ABSTRACT**

Artificial Intelligence (AI) has emerged as a revolutionary technology in the current healthcare setting that allows for efficient disease diagnosis, prediction, and patient management. This review article offers an extensive review of AI-based approaches like Machine Learning (ML) and Deep Learning (DL) for the diagnosis of significant diseases such as cancer, diabetes, cardiovascular diseases, neurological disorders, and infectious diseases. The article reviews research articles published on significant scientific portals that strictly adhere to the PRISMA protocol that focuses on image data, Electronic Health Records (EHER), genomics information, and wearable sensor information. Other performance parameters like accuracy, sensitivity, specificity, Area Under the Curve (AUC), precision, recall, and F1 measure have been discussed for an understanding of the effectiveness of the approach. The article

also focuses on the use of AI-based smart healthcare solutions and Internet of Things (IoT)-related devices that monitor diseases on a real-time basis. Nonetheless, despite the significant progress that AI has shown, issues of data privacy and the scarcity of data pose challenges.

**KEYWORDS:** *Artificial Intelligence; Machine Learning; Deep Learning; Disease Diagnosis; Predictive Healthcare; Medical Imaging; Clinical Decision Support Systems;*

*Pharmaceutical Research; Smart Healthcare Systems; Internet of Things (IoT).*

## INTRODUCTION

Artificial Intelligence (AI) is one of the most revolutionary technologies in contemporary healthcare science, especially in the diagnosis of diseases. AI has made it possible to analyze large amounts of data in the medical field with the use of computers and has made significant inroads in the diagnosis of various diseases. Among various diseases that are being diagnosed through AI innovation, the diagnosis of cancer, diabetes mellitus, and cardiovascular diseases is most notable. Not only is this diagnosis prevalent in almost every corner of the world, but this disease has high mortality rates as well.

Early diagnosis is essential for an improvement in patients in addition to providing good treatment at low costs. AI has made significant innovations in this area in addition to providing ease in diagnosis.

Cancer, diabetes, and heart conditions account for a large proportion of morbidity and mortality across the globe. Cancer is still a leading cause of death and is estimated to have millions of new cases every year. Diabetes is a chronic illness and impacts hundreds of millions of people and is a predisposing cause for heart conditions, renal failure, and blindness. Heart conditions, ranging from coronary heart disease and heart failure, are still the leading causes of mortality across the world. The rising incidence of such conditions and a lack of healthcare personnel make a tremendous need to have intelligent and scalable diagnostic systems imperative. AI systems are increasingly being looked at to address this challenge.

Essentially, AI in medical diagnosis involves methodologies like machine learning (ML), deep learning (DL), natural language processing (NLP), and computer vision. These methodologies make it possible for an algorithm to learn from past medical data and become smarter in the process with time, without requiring programmers to directly teach the computer how to accomplish all medical tasks.

In the medical domain, an AI system can analyze images, lab results, electronic health records (EHRs), genomic information, and even wearable data. Through the integration and interpretation of such broad-ranging information, an AI system can help medical professionals detect any patterns that could be hard and time-consuming in terms of

conventional methods.

Perhaps the most important strength that AI brings to diagnosis is that AI can handle massive amounts of information at incredible speeds. While human professionals possess immense talent, they also have limitations in terms of fatigue, human bias, and variability in human experience. On the other hand, AI systems have the ability to perform steadily at incredible speeds and thousands of points concurrently. It is an aspect that would prove to be extremely valuable in conditions such as cancer, diabetes, or cardiovascular disease, where minute changes in information may provide clues about the beginning or progression of the disease. Hence, AI not only assists health professionals but also helps them reach better diagnoses.

### **ARTIFICIAL INTELLIGENCE TECHNIQUES IN DISEASE DIAGNOSIS AND PREDICTION**

AI is a broad area integrated within many fields of mathematics and science. Any task that a machine can perform automatically, would be considered "intelligence" would be a subset of AI. AI algorithms are trained on population representation information. One of the most important subfields of AI is ML and the essential subfields of ML are Neural Networks and DL. The ML's objective is that the machine can train itself based on an input data set, experience, and receiving information from feedback. The ML algorithm optimizes itself with regard to the information received from the feedback in order to be as accurate as is possible in a particular task. Ideally, the ultimate goal is that it should work accurately on new unseen data sets as well. Imaging source is one of the most commonly used tools in the medical area for diagnostic patient information. Yet, this modality relies on human interpretation and is subject to increasing resource challenges. Automatic diagnosis of medical imaging by means of AI-in particular, in the field of DL-has effectively solved the problems of human error caused by inaccuracies or lack of sufficient experience. AI also plays an important role in image-based disease classifications, computer-aided diagnosis CAD, and image disease segmentation. As such, it is quite impossible to accurately get the tissues and organ images in the health system. While similar problems can be simulated with simple equations, medical imaging diagnosis tasks must be learned from experience. Continuous checking and reviewing of data are required for the detection of any disease and the prevention of its spread. Prompt Well-informed action has enormous social and financial implications for the lives of people worldwide. The application of AI in healthcare has enhanced the gathering and processing of useful data, and at higher levels, Programming of

surgical robots AI describes the power of a machine to study how a human learns by recognizing an image. And pattern recognition in a problematic situation. AI in health care has changed the way that information is collected, analyzed, And developed for patient care.

## **MACHINE LEARNING IN HEALTHCARE**

The major phases for developing a ML-based healthcare system are illustrated and major types of ML/DL that can be used in healthcare applications are briefly described next.

### **Un-Supervised Machine Learning**

The ML techniques utilizing unlabelled data are known as unsupervised learning methods. Widely used examples of unsupervised learning methods are a clustering of data points using a similarity metric and dimensionality reduction to project high dimensional data to lower-dimensional subspaces (sometimes also referred to as feature selection). In addition, unsupervised learning can be used for anomaly detection, e.g., clustering Classical examples of unsupervised learning methods in healthcare and prediction of hepatitis disease using principal component analysis (PCA) which is a dimensionality reduction technique.

### **Supervised Machine Learning**

Such methods that build or map the association between the inputs and outputs using labeled training data are characterized as supervised learning methods.<sup>[10]</sup> If the output is discrete then the task is called classification and for a continuous value output, the task is called regression. Classical examples of supervised learning methods in healthcare include the classification of different types of lung diseases (nodules) and recognition of different body organs from medical images. Sometimes, ML methods can be neither supervised nor unsupervised, i.e., where the training data contains both labeled and unlabelled samples. Methods utilizing such data are known as semi-supervised learning methods.

### **Semi-Supervised Machine Learning**

Semi-supervised learning methods are useful when both labelled and unlabelled samples are available for training, typically, a small amount of labelled data and a large amount of unlabelled data. Semi-supervised learning techniques can be particularly useful for a variety of healthcare applications as acquiring enough labelled data for model training is difficult in healthcare.

### **Reinforcement Learning**

Methods that learn a policy function given a set of observations, actions, and rewards in response to actions performed over time fall in the class of reinforcement learning (RL). RL has a great potential to transform many healthcare applications and recently, it has been used for context-aware symptoms checking for disease diagnosis.

### **APPLICATIONS OF MACHINE LEARNING IN HEALTHCARE**

Healthcare service providers generate a large amount of heterogeneous data and information daily, making it difficult for the "traditional methods" to analyze and process it. ML/DL methods help to effectively analyze this data for actionable insights. In addition, there are heterogeneous sources of data that can augment healthcare data such as genomics, medical data, data from social media, and environmental data, etc.

### **Applications of ML in Prognosis**

Prognosis is the process of predicting the expected development of a disease in clinical practice. It also includes identification of symptoms and signs related to a specific disease and whether they will become worse, improve, or remain stable over time and identification of potential associated health problems, complications, ability to perform routine activities, and the likelihood of survival. As in clinical setting, multi-modal patient's data is collected, e.g., phenotypic, genomic, proteomic, pathology tests results, and medical images, etc., which can empower the ML models to facilitate disease prognosis, diagnosis and treatment. For instance, ML models have been largely developed for the identification and classification of different types of cancers, e.g., brain tumor and lung nodules. However, the potential applications ML for disease prognosis, i.e., prediction of disease symptoms, risks, survivability, and recurrence have been exploited under recent translational research efforts that aim to enable personalized medicine. However, the field of personalized medicine is nascent that requires extensive development of adjacent fields like bioinformatics, strong validation strategies, and demonstrably robust applications of ML thus to achieve the huge and translational impact.

### **Applications of ML in Diagnosis Electronic Health Records**

Hospitals and other healthcare service providers are producing a large collection of electronic health records (EHRs) on a daily basis and comprise of structured and unstructured data that contains a complete medication history of patients. ML-based methods have been utilized for the extraction of clinical features for facilitating the diagnosis process.

### **ML in Medical Image Analysis**

In medical image analysis ML techniques are used for efficient and effective extraction of information from medical images that are acquired using different imaging modalities such as magnetic resonance imaging (MRI), computed tomography (CT), ultrasound, and positron emission tomography (PET), etc. These modalities provide important functional and anatomical information about different body organs and play a crucial role in the detection/localization and diagnosis of abnormalities. The key purpose of medical image analysis is to assist clinicians and radiologists for efficient diagnosis and prognosis of the diseases. The prominent tasks in medical image analysis include detection, classification, segmentation, retrieval reconstruction, and image registration.

### **DEEP LEARNING IN DISEASE DIAGNOSIS**

Modern healthcare advances because deep learning applications transform disease diagnosis, specifically in medical imaging analysis, laboratory diagnostic procedures, and predictive analysis systems. Effective disease detection happens through deep learning approaches, which produce accurate and quick diagnoses before symptoms appear, leading to superior patient results. Complex information recognition capabilities of artificial intelligence (AI) in large datasets form the basis of its role as a crucial medical tool in scientific research and clinical work.

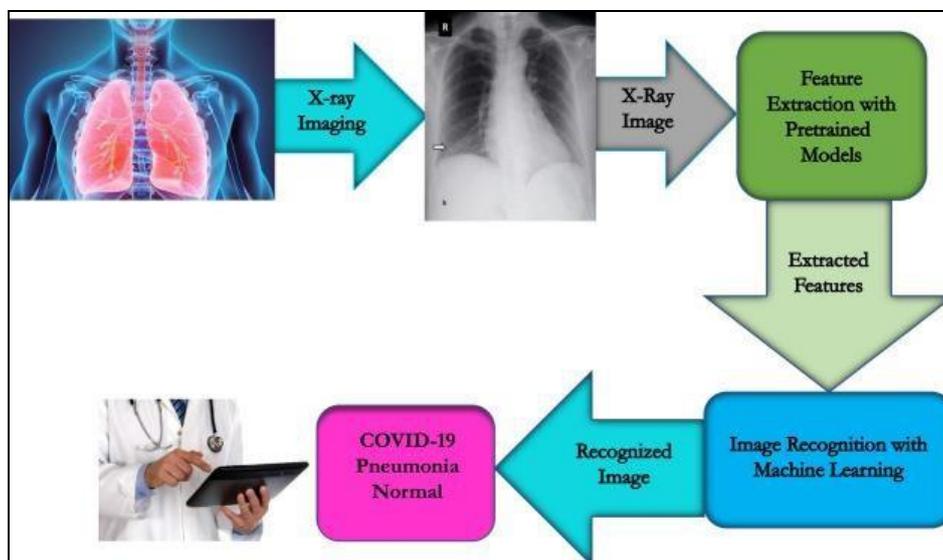
### **Medical Imaging and Pattern Recognition**

Medical imaging receives its most vital contribution through deep learning techniques in healthcare. Convolutional neural networks (CNNs) perform well when analyzing images from radiological, pathological, and dermatological medical studies.

These networks display superior abilities to find complex visual patterns in images, leading to highly precise disease detection and diagnosis abilities. X-ray, MRI, and CT scan interpretation benefit from their wide clinical use, enabling the detection of lung cancer, pneumonia, and brain tumors. By using vast datasets for training, these models become progressively more skilled in medical diagnostics tasks and surpass human capability.

Transformer-based models represent another significant advancement in medical imaging by providing effective tools for this purpose. Transformers excel over traditional CNN architectures because their design enables the identification of both distant relationships between image data and context-based information, which improves the quality of analysis in

complicated medical situations. Their medical use has expanded within radiology, producing better results for detecting abnormalities. When performing feature extraction, scientists use transformer design architecture to enhance breast cancer detection in mammograms to minimize false error rates.



**Figure 1: Advanced pattern recognition tools for disease diagnosis.**

Applying deep learning techniques produced major benefits for diagnosing dermatological and pathological conditions. Digital pathology AI systems inspect high-definition histopathological images to detect cancer cells at a very high-efficiency level. Deep learning technology serves dermatological purposes by analyzing skin lesions to distinguish benign and malignant conditions. The innovations have simultaneously decreased expert-related diagnostic uncertainties while boosting the identification of conditions in their early stages to enable appropriate medical treatments.

### **AI in Laboratory Diagnostics**

Deep learning has revolutionized laboratory diagnostic procedures through its ability to handle large biological data such as clinical blood results combined with histological exams and genomic information. The AI-powered systems help medical experts discover invisible patterns in patient data. Thus, they prevent misdiagnosis while streamlining medical work. Separate workflows powered by AI function better and deliver more precise results, therefore improving the identification of diseases and patient care.

Deep learning technology delivers substantial benefits through its laboratory diagnostic

utilization, specifically in hematology. AI models perform automated blood testing as part of disease detection systems to discover the hematological disorder known as leukemia. Evaluating blood smear pictures through deep learning systems allows precise diagnosis of leukemia types, subsequently enabling proper treatment customization. AI models observe cell structures and differentiate normal cells from abnormal ones to assist hematologists through their diagnostic process. Implementing artificial intelligence cuts down diagnosis time and minimizes the requirement for manual biological sample examination, thus enabling laboratories to handle more specimens quickly.

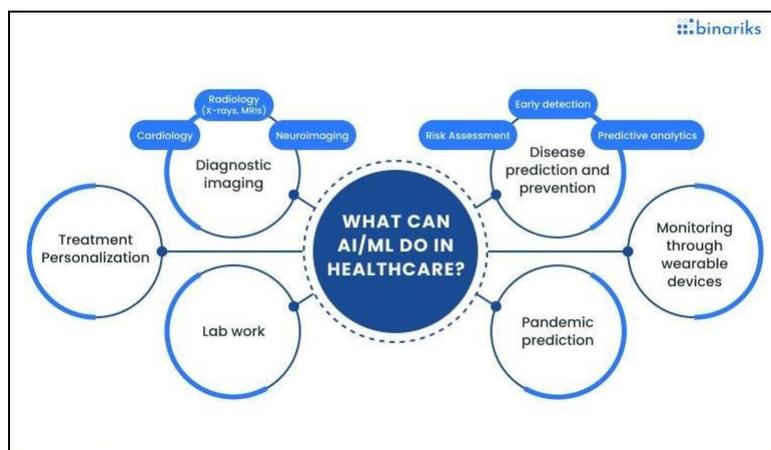
AI technology resulted in advanced biopsy examination capabilities as part of laboratory diagnostic systems. Pathologists formerly conducted manual assessments of tissue samples to search for cancer cells; however, this traditional approach takes too much time and contains space for human inaccuracies. Machine learning algorithms develop capabilities to inspect biopsy slides where they determine malignant conditions with notable precision while maintaining high accuracy levels. Identifying tiny cell anomalies improved with AI diagnostic tools results in accurate early condition detection for breast cancer and prostate cancer treatment.

Genomic sequencing benefits from deep learning technology through its significant knowledge development. AI software conducts large-scale assessments of genetic information to disclose the presence of hereditary disease-related mutations. Genomic profiling data through this discovery enables healthcare professionals to develop custom treatment plans to fit individual genetic makeup. Numerous medical conditions, including cardiovascular diseases and diabetes with their various cancer types, can be forecast through AI-based genetic marker evaluation technology.

Combining AI with genomic studies enables better-customized therapies, completely modernizing present disease prevention practices.

### **Early Disease Detection and Predictive Analytics**

The most outstanding healthcare benefit of deep learning involves its predictive power to detect diseases before their symptoms become apparent. AI systems read medical records and genetic risks to estimate disease potential within an individual. Through deep analysis of vast data supplies, these learning models reveal delicate patterns that point to preliminary disease development so medical staff can take early action, leading to superior health results.



**Figure 2: AI/ML Algorithms for Early Disease Detection and Diagnosis.**

Neurodegenerative disorders such as Alzheimer's disease present significant challenges in early diagnosis. Doctors recognize disease only when the patient starts showing cognitive difficulties, interfering with treatment success. Research shows deep learning models produce outstanding results when identifying Alzheimer's disease development through scanning neurological data. AI analyzes MRI images to identify preliminary disease indicators through brain atrophy distribution measurements and monitoring changes in the brain network connections. The prediction systems allow medical professionals to start necessary treatments early, thus matching disease progression and improving patient quality of life.

Diabetes patient care has progressed through the development of AI-based predictive analytical systems. Deep learning algorithms base their blood sugar level change predictions on continuous glucose monitoring data and other metabolic indicators to enhance patients' condition-tracking abilities. Doctors can generate specific exercise and dietary advice through AI applications, strengthen insulin usage, and prevent diabetic complications. The exact approach to disease management enables patients to follow treatment plans better, resulting in better health outcomes.

The detection, along with the containment of infectious disease outbreaks, becomes possible because of AI. AI models with deep learning capabilities analyzed public health information, human movements, and virus genetics to forecast pandemic developments throughout the COVID-19 crisis. Implementing AI surveillance helped public health technicians execute timely disease containment procedures, which slowed disease transmission. The predictive analytics systems remain beneficial because they help identify new infectious threats while enabling quick response planning and mitigation operations.

Medicine has advanced disease diagnosis through deep learning integration because these techniques improve clinical imaging, laboratory detection methods, and predictive analysis systems throughout healthcare. Implementing AI-driven solutions improves medical detection times by creating enhanced treatment strategies that lead to superior patient health results and active healthcare provision. Deep learning research developments will expand the disease diagnosis revolution until artificial intelligence influences medical choices.

## **1. AI and ML in Cancer Diagnosis**

### **Application in Imaging**

The field of medical imaging has achieved a lot in AI especially in early detection of cancer. Based on deep learning algorithms, AI systems can process medical images like mammogram, CT scans and MRIs to detect early cancer. These algorithms are trained on large bodies of annotated images which enables them to identify patterns that are usually intangible to the human radiologists. As an illustration, AI has demonstrated superior precision in mammograms to diagnose breast cancer compared to human experts in both sensitivity and specificity. On the same note, AI uses in the detection of lung cancer have proved efficient in the detection of nodules in the chest X-rays and CT scan resulting in an earlier intervention and better outcome.

### **Genomic Data Analysis**

Another aspect in which machine learning is changing the cancer diagnosis process is the examination of genomic data. Using the genetic makeup of cancer cells, the ML models are able to detect the mutations and biomarkers that are indicative of the occurrence of a particular type of cancer. Genomic testing can be used as an example, in which patients that are in danger of cancer like, breast or colorectal cancer can be identified to undergo screening and detecting early cessation. Furthermore, AI-based approach that makes use of genomic information can develop individual treatment prescriptions depending on the genetic makeup of a patient and help assess the effectiveness of treatment and minimize the requirements of unwanted drug reactions.

### **Case Studies and Clinical Trials**

A variety of clinical trials and case studies have confirmed the usefulness of AI in cancer diagnosis. In a study involving deep learning models to interpret mammograms done by Google Health, it was discovered that AI had the potential to reduce false negatives and false

positives and increase the overall accuracy of breast cancer screening. On the same note, AI algorithms have been used in the diagnosis of lung cancer and researches have indicated that AI-based tools are more effective in the detection of lung cancer nodules compared to the conventional ones. Those are already tested in clinical institutions and with good results demonstrating that AI can transform the concept of early cancer diagnosis.

## **2. Artificial Intelligence and Machine Learning in Diabetes detection: Predictive Modeling**

Predictions of the occurrence of diabetes (especially type 2 diabetes) are increasingly being performed with the use of machine learning algorithms. ML models are able to determine the risk of an individual developing the disease by examining a number of factors, which may include age, sex, family history, lifestyle, and clinical history. Lifestyle changes like eating habits and physical exercises can prevent or slow the development of diabetes because of early prediction. As an illustration, we can see that ML has been used to predict diabetes in the creation of risk calculators, including the Finnish Diabetes Risk Score (FINDRISC) that uses given risk factors to estimate the likelihood of an individual developing diabetes.

### **Continuous Monitoring**

The use of AI-interested blood glucose level monitoring tools to provide continuous monitoring is changing the way diabetes is managed. Glucose analyses are more regularly being carried out by wearable devices, which operate in a manner that uses AI algorithms to assess the glucose levels in real-time. The gadgets also notify the sufferers when their sugar levels are not in the optimal scope, and thus they take the necessary remedial measures before the complications set in. Moreover, AI programs can also forecast blood sugar levels changes and help patients regulate the dose of insulin a lot more precisely and prevent life-threatening swings.

### **Influence on Early Intervention.**

AI and ML technologies can greatly enhance the results of patients since they can be diagnosed with diabetes early enough and therefore timely interventions can be taken. As an example, algorithms based on artificial intelligence can foresee the risk of diabetic complications, including neuropathy and retinopathy to implement preventive strategies early. Also, prediabetes detection early in life can result in lifestyle emendations, which revert the condition to non-diabetes state before it develops to full-blown diabetes. This pro-

active measure enhances quality of life as well as a decrease long-term healthcare costs, incurred because of diabetes-related complications.

### **3. AI and ML in Heart Disease Diagnosis**

#### **Risk Stratification Models**

Risk stratification models on heart disease are being developed using AI and ML. These models are used to measure various cardiovascular risk factors (including blood pressure, smoking state, cholesterol, and family history) to determine the risk of heart disease occurrence. Through the analysis of the great numbers of data in electronic health records (EHRs), AI systems can choose those individuals who are at the risk of having heart disease in order to intervene in time. An example of such applications is the prediction of the onset of a heart attack after the abnormalities are detected by evaluating trends within electrocardiogram (ECG) measurements, which can signal possible cardiovascular disease.

#### **Embedded technology: Wearable computing**

Artificial intelligence is improving the diagnosis and treatment of heart diseases by combining the wearable technology with AI. Smart devices such as smartwatches and fitness trackers to track the heart rate, blood pressure, and physical activity rates are gathering enormous amounts of data, and their data can be analyzed in real-time with AI algorithms. Such devices are able to detect abnormal heart beats, including atrial fibrillation, and notify the intended users to consult a doctor before the condition escalates to a more serious condition, including stroke or heart attack. Such combination of constant monitoring and AI analysis could result in the early detection and prompt treatment of the malady and decrease the risk of heart attack.

#### **Clinical Implementation and Outcomes**

Research has also indicated that AI largely affects the diagnosis of heart diseases. To illustrate the point, AI algorithms used in ECG data have been shown to recognize arrhythmias with a high level of accuracy and could help avoid life-threatening scenarios. Also, AI applications are being applied to the imaging data in the form of echocardiograms or coronary angiograms to identify the signs of coronary artery disease to further help in early detection and treatment.

### **4. Comparative Analysis**

#### **Comparison between AI/ML Approaches in Diseases**

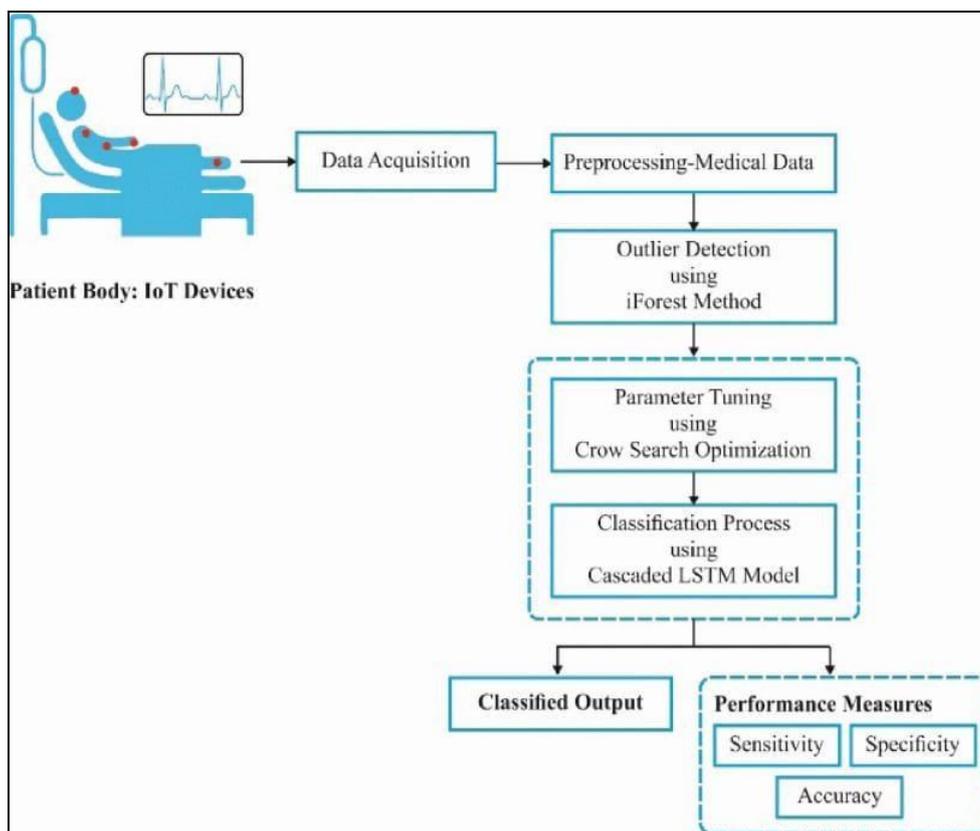
Although the applications of AI and ML are different in the case of cancer, diabetes, and heart diseases diagnostics, the similarities are multiple in terms of the application of these

technologies. The use of AI and ML models in reviewing large datasets, which could be imaging, genetic or clinical data, detects the disease early and lessons on its future occurrences in all three diseases. Moreover, AI and ML personalize the treatment plan, which the care delivered to an individual patient. Nevertheless, every disease is a challenge when it comes to diagnosis. As an example, in cancer diagnosis, AI is involved mostly in image-based and genomics analysis, whereas in diabetes, it is more based on predictive modeling and continuous measurement. In heart disease the AI is mostly utilized in risk evaluation and bendable technology incorporation.

### **Challenges and Limitations**

Although the advantages of AI and ML in disease evaluation are willing, the system encounters a number of challenges. The question of privacy of data is also a major concern as the personal health data is being used, casting doubts on consent and security. Another issue is algorithmic bias, with AI systems being able to be only as good as the data they are trained on, and a biased dataset may result in biased output. Additionally, AI models do not always require small or heterogeneous datasets to be trained effectively and this can be hard to get, particularly in cases of underrepresented groups of individuals.

Overall working process. The proposed methodology is pretty power-efficient considering the previous wireless communications with high freedom of activity in case of external movements of the users. Furthermore, small, low-weight IoT devices are utilised in the presented model, which are user-friendly. Some such IoT devices include smartphones, wrist-band, smartwatch and so forth.



**Fig 3: The Proposed Smart Healthcare Diagnosis Model.**

The embedded sensors are used to proceed with heavy computations in order to estimate and identify normal and abnormal heart rates. Smart devices for instance smartphones are embedded to the subjects, which can be taken anywhere in pockets. Moreover, embedded ECG and temperature sensors are highly recommended in order to gather data regarding heart parameters of the subject. From this data, the results of their common life style also can be identified. When the data is received through low-power Bluetooth communication, smartphones process the data and categorize it as healthy/unhealthy. The android platform carries out the prediction of diabetes and efficient heart rate. IoT devices collect the patient data initially and preprocess it in order to transform the data into compatible format. Pre-processing includes a few steps such as transformation of data, conversion of formats, and labeling of classes. Then, iForest technique is employed in order to get rid of outliers exist in the patient data. Followed by, CSO-CLSTM model is used in order to classify the data into existence and non-existence of the disease.

## CONCLUSION

This book review focuses on the important use of artificial intelligence and machine learning to change the present scenario of disease diagnosis, prediction, and overall healthcare

delivery. AI- powered machine learning algorithms, including supervised learning, unsupervised learning, deep learning algorithms, and many more, have proved to be very effective and fruitful in diagnosing severe healthcare conditions like cancer, diabetes, cardiovascular disease, neurological disorders, and infective diseases. Also, the use of AI-powered healthcare monitoring systems with medical imaging, EHRs, genomic information, and wearable sensor systems has proved to be very effective and fruitful in monitoring patients proactively and reducing the burden of the hospital.

Although tremendous progress has been made in AI-based healthcare systems, there are still certain obstacles that restrict its extensive implementation in clinical settings. These obstacles include privacy issues in healthcare data, absence of a universal healthcare dataset, bias in AI algorithms, and a requirement for healthcare images and clinical labels in large quantities. Moreover, explainability and clinical validation of AI-based systems are also essential to gain acceptance among healthcare professionals in the future. AI has tremendous potential to increase diagnosis accuracy, decrease healthcare costs, and provide better patient outcomes and is an integral part of next-generation intelligent healthcare systems.

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