



Machine Learning for Early Disease Diagnosis: A Review of Techniques in Healthcare Applications

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ABSTRACT

Early disease detection has long been a cornerstone of healthcare, with the adage “prevention is better than cure.” The rise of machine learning (ML) has revolutionized this field, enabling the analysis of vast medical data to predict health issues before they become clinically evident. Traditional detection methods, relying on manual examinations and patient history, are often limited by human error and subjectivity, especially as medical datasets grow in size and complexity. This review addresses the gap between conventional diagnostic methods and ML’s potential in early disease detection. It explores various ML algorithms used across domains such as heart disease, diabetes, and cancer, and tracks the evolution of ML techniques from 2015 to 2024. The review also examines ethical and technical challenges, particularly concerning data privacy. Emerging trends in the field, such as the integration of ensemble learning and deep learning models, are also proposed. The significance of this work lies in its comprehensive overview of ML in early disease detection, highlighting its transformative potential in improving diagnosis accuracy, reducing healthcare costs, and enhancing patient outcomes.

Keywords: Machine learning, Early disease detection, Healthcare applications, Deep learning, Data privacy

Introduction

Throughout history, healthcare has been guided by the aphorism “prevention is better than cure.” Early detection is not only better for the patient but also a huge drop in the strain on the healthcare system. This has led us into a new era of predictive potential for early disease detection via machine learning (ML), which has revolutionized how early disease detection is taking place, while the mainstays of early disease diagnosis have in the past relied on manual examination, patient history, and simple diagnostic tools. Physicians have long been taught to identify trends and departures from the norm and to base their choices on their clinical expertise. However, because they rely on human judgment, which is subjective and prone to error, these approaches are frequently constrained.¹ Additionally, the vast and disjointed datasets produced by modern medicine may be too big for conventional approaches to handle. A rapidly evolving subset of artificial intelligence (AI), the ability to create algorithms that can look at data, pattern them, and predict future events is intriguing. ML techniques are a rapidly growing trend for early detection of diseases because they can process and analyze massive amounts of data, or “big data.” Because of this change in strategy, we have been able to build prediction models, along which patterns and connections that are potentially too subtle for us to pick up can now be learned and perceived.²

At the same time, one of the main advantages of ML in early disease diagnosis is that it can treat a huge number of factors simultaneously. For instance, ML algorithms can take many more inputs such as genetics, family history, and other personal behaviors and environmental situations, whereas traditional approaches might only look at a few different symptoms. An advantage of this method is that it allows for the measurement of illness risk more precisely and individually for each patient. ML approaches have been found to add value in a range of medical specialties. For example, in radiology, sophisticated imaging methods including computed tomography (CT) scans and magnetic resonance imaging (MRI) produce tens of millions of pixels of data that will surpass the capacity of the human radiologist to view in their entirety.³ ML algorithms can quickly filter through these photos, and automatically identify very small irregularities which the human eye may not detect. In much the same way, we are also using ML algorithms to improve our ability to analyze DNA sequences to find hereditary susceptibilities to disease. One approach to healthcare is iterative ML. The more data they can process, the better their algorithms tend to perform. Early disease identification is one place where this adaptability is especially helpful given that symptoms are often very complex and highly dynamic. ML algorithms have the tendency to increase the accuracy with which they can detect early illness markers when continuing learning and improvement in prediction.⁴

However, incorporating ML into early disease diagnosis is not without its challenges. Because medical data is sensitive, worries about data security and privacy continue to be the most pressing issues. Making sure ML models are visible and explicable is a second challenge; certain algorithms are “black boxes,” which could prevent their clinical use. Furthermore, because historical data is not entirely equitable, it can skew healthcare outcomes for the patients who get it. Compared to traditional methods, ML for early disease diagnosis represents a paradigm change in healthcare. The fundamentals are covered by traditional approaches, but ML capabilities enable previously unattainable scales for processing large amounts of data and deriving useful insights. In order to effectively leverage the predictive power for early disease diagnosis and promote healthy populations and healthcare systems, medical specialists and data scientists will need to collaborate as technology continues to advance.⁵

The aim of this study is to explore the role of ML, ensemble learning (EL), and deep learning (DL) models in early disease detection, analyzing their evolution, common applications, and performance

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across various diseases. The study also examines the ethical and technical challenges associated with these technologies, particularly concerning data privacy, while proposing emerging trends for future advancements in the field. The contributions of the study are stated below:

- i. Identification of common ML, EL, and DL models used in early disease prediction.
- ii. Analysis of the evolution of these models, along with an exploration of challenges and emerging trends in the field.

The rest of the article is organized as follows: Section 2 presents the architecture of the early disease prediction system, detailing the components and workflow. Section 3 explores various ML techniques applied to different disease diagnoses, highlighting their effectiveness. Section 4 provides a discussion of the findings and implications of the proposed methods. In Section 5, the challenges and opportunities in early disease detection are addressed, focusing on current limitations and potential advancements. Section 6 offers a glimpse into the future, discussing emerging trends and prospects in ML-driven early disease diagnostics. Finally, Section 7 concludes the article with a summary of key insights and future directions.

Architecture of the Early Disease Prediction

The process starts with collecting medical datasets derived from various sources, including patient data and

electronic health records. These raw datasets undergo preprocessing to clean, normalize, and organize the information, ensuring consistency and accuracy. Following this, feature extraction and selection are performed to identify and prioritize the most relevant features or variables for disease prediction, reducing dimensionality and improving computational efficiency.

Once the data is prepared, advanced computational methods are applied, including DL, ML, and optimization-based techniques. These methods analyze the data to identify patterns and relationships indicative of disease risks. The performance of these models is then evaluated using metrics like accuracy, precision, F-measure, and processing time. Based on this evaluation, the best-performing model is selected and used for disease prediction. The ultimate goal of this architecture is to facilitate accurate and efficient early detection of diseases, enabling timely medical intervention. The general architecture of early disease prediction is shown in Figure 1.

Machine Learning Techniques for Different Disease Diagnoses

Several authors and scholars have employed ML methods to diagnose illnesses. This section explains the significance and influence of different forms of ML-based disease diagnostics (MLBDD) that have been debated extensively. For example, as COVID-19 is a worldwide problem, a lot of research publications from 2020 to the present have concentrated on utilizing ML to diagnose COVID-19 disease, which is what we also gave

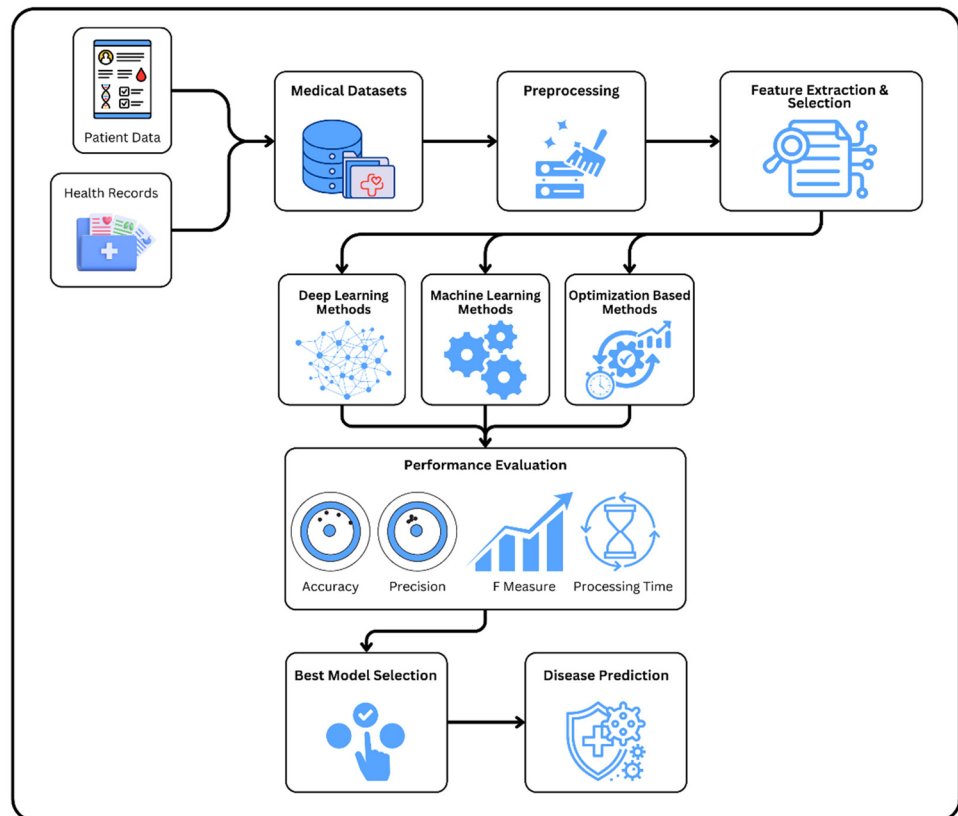


Fig 1 | General architecture of early disease prediction

priority to in this study. Heart disease, kidney disease, breast cancer, diabetes, Parkinson’s, Alzheimer’s , and COVID-19 are among the serious diseases; additional illnesses are briefly covered under the “Other Diseases” heading.

Heart Disease

ML algorithms are used by most researchers and experts to detect heart problems.^{6,7} For example, Ansari et al.⁶ introduced an automated method for diagnosing coronary heart disease based on neuro-fuzzy integrated systems, which achieved an accuracy of about 89%. The absence of a thorough description of how the suggested approach might be applied in numerous situations, such as multiclass classification, enormous data processing, and unequal distribution of class, is one of the study’s main flaws. Additionally, the model’s prediction validity is uncertain, which has recently raised serious concerns in the medical community, particularly for patients who are unfamiliar with medical terminology. In order to identify irregular heartbeats, Rubin et al.⁸ used a deep convolutional neural network (CNN). The researchers of the study subsequently modified the loss function as a strategy to increase the training set’s sensitivity and improve the specificity. They submitted their model to the PhysioNet classification computer competition in 2016. With an ultimate forecast of 0.95 specificity and 0.73 sensitivity, they place second in the competition.

In addition, algorithms based on the DL have been recently employed to identify heart disease. For example, Miao and Miao⁹ presented a DL method for determining electronic fetal monitoring (EFM) based on the multiclass morphologic pattern. The model identifies the morphologic pattern of patients with pregnancy problems. An F1 score of 0.85, an accuracy of 88.02% and a precision of 85.01% are some additional preliminary

computational findings. The morphologic pattern of patients with pregnancy problems is identified using the model. Additional preliminary computational findings include an F-score of 0.85, an accuracy of 88.02%, and a precision of 85.01%. They acknowledged that a greater accuracy rate was a benefit of using a variety of dropout techniques to address the overfitting problems in that study, which also improved training time.

Khader Basha et al¹⁰ utilized a hybrid ML approach combining decision tree (DT) and AdaBoost to enhance coronary heart disease prediction, focusing on cardiac and kidney disorders. Their model’s performance was measured by accuracy, true positive rate (TPR), and Specificity. Meanwhile, Chandrasekhar and Peddakrishna¹¹ tested six algorithms (random forest, K-nearest neighbor, logistic regression (LR), Naïve Bayes, gradient boosting, and AdaBoost) on Cleveland and IEEE DataPort datasets. Using GridSearchCV and fivefold cross-validation, LR achieved 90.16% accuracy on Cleveland, while AdaBoost reached 90% on IEEE DataPort. Their soft voting ensemble classifier further improved accuracy to 93.44% (Cleveland) and 95% (IEEE), outperforming individual models and prior studies.

Although there is a wealth of research on using ML to diagnose cardiac disease,¹² none of it has addressed the difficulties associated with multiclass categorization of unbalanced data. In most situations, nevertheless, the model’s capacity to explain the ultimate forecast is severely constrained. Some of the cited studies that employed ML and DL to identify heart disease are shown in Table 1 of this article.

The studies presented focus on various ML algorithms applied to heart disease prediction using datasets like Cleveland and MIT-BIH. Key findings reveal that random forest (RF) and support vector machine (SVM) models consistently perform well, with RF achieving

Table 1 | Comparison of Machine Learning Techniques in Heart Disease Detection

Ref	Year	Algorithms Utilized	Dataset Used	Data Format	Performance Metrics
13	2021	Gaussian Naïve Bayes, Bernoulli Naïve Bayes, RF	Cleveland dataset	Tabular	Gaussian Naïve Bayes achieved an accuracy of 85.00%.
14	2020	RF, CNN	Cleveland dataset	Tabular	Random forest achieved an accuracy of 80.33%.
15	2019	Support vector machine (SVM)	Cleveland database	Tabular	Accuracy observed across various methods ranged from 73% to 91%.
16	2019	Back-propagation neural network, logistic regression	Cleveland dataset	Tabular	BNN achieved an accuracy of 85.07%, while logistic regression achieved 92.58%.
17	2018	SVM, Cuckoo Search-Optimized Neural Network	Cleveland dataset	Tabular	Support vector machine achieved an accuracy of 94.44%.
18	2017	CNN	MIT-BIH dataset	Tabular	For balanced data, accuracy was 94%, whereas for imbalanced data, accuracy dropped to 89.07%.
19	2018	SVM	MIT-BIH dataset	Tabular	Imbalanced data analysis yielded 97.77% accuracy, while noise-free ECGs achieved 97.08% accuracy.
10	2023	Decision tree, AdaBoost	Cleveland	Tabular	AdaBoost outperformed decision tree.
11	2023	RF, KNN, logistic regression, Naïve Bayes, gradient boosting, AdaBoost, soft voting ensemble	Cleveland, IEEE DataPort	Tabular	Logistic regression achieved an accuracy of 90.16%, while AdaBoost also achieved 90%.

accuracies around 80%–85% and SVM reaching up to 94.44%. In some cases, ensemble methods like Adaboost and soft voting ensemble showed competitive results, with LR achieving 90.16%. Back-propagation neural networks and CNNs also performed strongly, with accuracy rates ranging from 89% to 97%, depending on data balance and noise levels. Overall, the studies highlight the effectiveness of both traditional and advanced ML techniques for heart disease prediction.

Kidney Disease

Renal disease commonly referred to as kidney disease is nephropathy or kidney damage. Kidney disease patients have a reduced ability of kidney function, and if left untreated it can cause kidney failure. The National Kidney Foundation estimates that 10% of the global population is affected by chronic kidney disease (CKD) and millions of people die annually because of inadequate care. Recent renal disease detection methods based on ML and DL may provide access to nations that cannot perform kidney disease diagnostic tests.²⁰ In order to assess four distinct ML methods, Charleonnann et al.²¹ employed publicly accessible datasets: Using LR, SVM, K-nearest neighbors (KNN), and DT classifiers, the datasets were classified; the corresponding accuracy rates were 96.55%, 98.3%, 98.1%, and 94.8%. Aljaaf et al.²² compared RPART, SVM, LOGR, and MLP with the help of the same dataset as CKD used by Charleonnann et al.²¹ and observed that MLP is the best model that can identify CKD with 98.1% accuracy. In order to detect prolonged kidney illness, Ma et al.²³ use datasets that contain records from numerous sources. They discovered that the accuracy of their proposed heterogeneous modified artificial neural network (HMANN) model ranged from 87% to 99%. Table 2 summarizes a few studies that employed ML and DL to identify kidney illness.

Islam et al.²⁴ explored the ability to use ML approaches to diagnose CKD and found that predictive modeling would greatly improve timely detection. In evaluating 12 ML classifiers, they concluded that XGBoost performed the best with accuracy, precision,

recall, and F1-score, all reaching 0.98. Furthermore, Sawhney et al.²⁵ developed a DL-based multi-layer perceptron classifier for CKD diagnosis, with 100% accuracy by using data from 400 patients. Using deep neural networks (DNN) from the PyTorch library, their approach surpassed conventional models like SVM and Naïve Bayes and proved that neural models can be used to handle nonlinear data and enhance diagnosis accuracy.

The studies on kidney disease detection using ML techniques show impressive results across various algorithms and datasets. RF and multi-layer perceptron (MLP) achieved perfect accuracy (100%) in some studies, while other models like feedforward neural networks, XGBoost, and artificial neural networks (ANN) reported high performance with accuracies above 95%. CNNs, used on image datasets, also demonstrated strong results, with accuracies ranging from 95% to 99.61%. Additionally, hybrid models like CNN-SVM and kernel clustering techniques enhanced detection accuracy and sensitivity. Overall, ML techniques, particularly ensemble methods and DL, exhibit robust potential for kidney disease detection.

Breast Cancer

ML has been proposed by a number of medical researchers as a means of detecting breast cancer in its early stages. For instance, Miranda and Felipe³¹ developed fuzzy-logic-based computer-assisted identification techniques for breast cancer classification. Fuzzy logic is superior to other traditional ML techniques because it can replicate the expert radiologist's method and way of thinking while lowering computational costs. If the operator enters factors like contour, form, and density, the algorithm provides a classification of cancer depending on the user's selection. The accuracy was roughly 83.34% based on the model that Miranda and Felipe³¹ proposed. The experiment's precision and fairness were improved by the authors' usage of almost an equal ratio of photos. However, since the authors did not explore how the results were explained, it is possible to claim that the results among benign and malignant

Table 2 | Comparison of Machine Learning Techniques in Kidney Disease Detection

Study	Year	Algorithms Utilized	Dataset Used	Data Format	Performance Metrics
26	2021	Naïve Bayes, decision tree, RF	Chronic kidney disease dataset	Tabular	Random forest achieved an accuracy of 100%.
27	2020	Artificial neural network (c), kernel K-means clustering	100 patient ultrasound images	Image	ANN achieved an accuracy of 99.61%.
28	2018	Logistic regression, feedforward neural network, wide DL	Chronic kidney disease dataset	Tabular	Feedforward neural network attained an F1-score of 99%.
29	2020	Convolutional neural network-support vector machine (CNN-SVM)	Proprietary dataset	Tabular	Accuracy of 97.67% was accompanied by a sensitivity of 97.5% and specificity of 97.83%.
30	2019	CNN	Proprietary dataset	Image	CNN achieved an accuracy of 95%.
24	2023	XGBoost	CKD dataset	Tabular	XGBoost attained an accuracy of 98.3% with an F1-score of 98%.
25	2023	MLP	400 patient data	Tabular	MLP achieved an accuracy of 100%.

classifications is accurate in general. Moreover, there is no confusion matrix which shows the actual prediction of models for each of the given classes.

Using SVM and k-means clustering (KMC), Zheng et al.³² suggested hybrid methods for identifying breast cancer. Using the Wisconsin Diagnostic Breast Cancer (WDBC) dataset, their suggested model achieved a 97.38% accuracy rate and significantly resolved the dimensional problem. The dataset has 32 variables organized into 10 categories and is regularly distributed. Applying their suggested model to a dataset with an uneven class distribution, where some items may be missing, makes it difficult to say that it will perform better.

Asri et al.³³ employed a number of ML models, like SVM, DT (C4.5), NB, and KNN, with the WBC datasets in order to categorize the best ML models. With an accuracy of 97.13%, the authors of the research demonstrated that SVM outperformed all other ML methods. However, the outcomes might be different if that same experiment is conducted on a different database. Furthermore, combining experimental findings from this work with ground truth values can provide a more precise indicator of the efficacy of a certain ML model.

Mohammed et al.³⁴ conducted an extremely similar investigation. In order to identify the most effective ML techniques, the authors used three algorithms: Classifiers are DT(J48), NB, and sequential minimal optimization (SMO). The data was gathered from two mostly used sets of data: WBC and breast cancer datasets. The second part is also interesting because the authors devoted special efforts to address the data imbalance problem and used data resampling labeling to alleviate it. However, their results have shown that the SMO algorithms perform better than the rest of the two classifiers to reach over 95% precision in both datasets. However, many resampling methods were used to balance the imbalance ratio, so the probability of data variety may be also affected. As such, these

three ML techniques have poor use when used on an uneven or non-normally distributed dataset. Assegie³⁵ used the grid search method to obtain the ideal KNN model parameters. They then found out that the performance of the model was greatly affected by the parameter tuning. With a little tweaking, they found that we could get close to 94.35% accuracy, as opposed to the default KNN of around 90%.

Bhattacharjee et al.³⁶ used a backpropagation neural network (BNN) to identify breast cancer. The WBC dataset with nine attributes yielded 99.27% accuracy. Alshayegi et al.³⁷ developed a shallow ANN to differentiate among types of breast cancer tumors using the WBCD and WDBI datasets. In doing so, the researchers showed that the proposed model could detect cancers with 99.85% accuracy without changing the algorithms or choosing characteristics.

Sultana et al.³⁸ used a different ANN architecture to detect breast cancer with the WBC dataset. The neural network (NN) models employed were modular neural networks (MNN), generalized feedforward neural networks (GFFNN), self-organizing feature maps (SOFM), multilayer perceptrons (MLP) neural networks, Jordan-Elman neural networks, recurrent neural networks (RNN), SVM neural networks, and probabilistic neural networks (PNN). According to their most recent computational results, the PNN outperformed the other neural network models utilized in that study with an accuracy of 98.24%. However, in contrast to many other studies this work does not establish which traits are most important when it makes its predictions, making it less interpretable.

DL was also employed by Ghosh et al.³⁹ In addition, the authors trained seven DL models, i.e. ANN, CNN, GRU, LSTM, MLP, PNN, and RNN, on the WBC dataset. We found that LSTM and GRU had the best performance out of all the DL models with accuracy reaching almost 99%. A selection of studies that employs ML

Table 3 | Comparison of Machine Learning Techniques in Breast Cancer Detection

Study	Year	Algorithms Utilized	Dataset Used	Data Format	Performance Metrics
42	2020	Naïve Bayes, Bayesian network, RF, decision tree (C4.5)	BCSC	Image	Bayesian network achieved an ROC of 0.937.
43	2015	SVM	Mini-MIAS, INBreast	Image	Mini-MIAS dataset results showed an accuracy of 99% with an AUC of 0.9325.
44	2015	SVM	IRMA, DDSM	Image	IRMA dataset achieved a sensitivity and specificity of 99%.
45	2017	Logistic regression-ANN	156 proprietary cases	Image	DDSM dataset achieved a sensitivity of 97% and specificity of 96%.
46	2016	Binary logistic regression	18 proprietary cases	Image	Binary logistic regression achieved an accuracy of 81.8% with an AUC of 0.855.
47	2015	Naïve Bayes, logistic regression with AdaBoost	246 proprietary images	Image	Logistic regression with AdaBoost achieved an accuracy of 80.39%.
40	2024	MLISBCP (K-Means SMOTE, Boruta)	Breast cancer dataset	Tabular	Proposed model reported sensitivity of 90% and specificity of 97.5%.
41	2023	KNN (with PCA/SVD)	Fine needle aspiration dataset	Tabular	KNN attained an accuracy of 97.53%.

and DL techniques to diagnose breast cancer is included in Table 3 below.

The risk of breast cancer is growing at an alarming rate, and Das et al.⁴⁰ proposed an intelligent system, MLISBCP, to enhance breast cancer prediction. Class imbalance handling was done by using K-Means SMOTE and feature selection was done using Boruta in the system, with an accuracy of 97.53%, better than the existing models. Furthermore, Shafique et al.⁴¹ concentrated on feature selection approaches to improve the prediction accuracy of breast cancer. In this regard, they had methods such as principal component analysis (PCA), singular vector decomposition (SVD), and chi-square, and tried several classifiers. However, the results of the group showed that using 15 features selected by PCA, KNN achieved perfect accuracy, demonstrating the promise of optimized feature sets and balanced datasets in enhancing prediction performance.

The studies on breast cancer detection using ML techniques demonstrate diverse approaches and strong performance across different datasets and algorithms. Models such as SVM achieved high accuracies, with the Mini-MIAS dataset showing 99% accuracy and an AUC of 0.9325. Hybrid models like LR combined with ANN and Naïve Bayes with AdaBoost achieved high sensitivity and specificity, with LR-ANN reaching 97% sensitivity and 96% specificity. The use of ensemble methods, like K-Means SMOTE with Boruta and KNN with PCA/SVD, also showed robust results, with sensitivity as high as 90% and specificity of 97.5%. Overall, ML models exhibit excellent potential for breast cancer detection, particularly with image and tabular data.

Diabetes

The IDF estimates that more than 382 million people around the world have diabetes at the current moment and this number may rise to 629 million by 2045.⁴⁸ Many studies have described the use of ML-based systems for identifying diabetes patients. For instance, Kandhasamy and Balamurali⁴⁹ investigated the performance of the following ML classifiers for diagnosing patients with diabetes mellitus, namely J48 DT, KNN, RF, and SVM. The investigation was performed on the UCI diabetes dataset and KNN classifier with $K = 1$, and RF classifier performed with almost perfect accuracy. Nevertheless, there is one drawback in this work: the employed diabetes dataset was binary and had only eight attributes. Therefore, it is not a surprise that you get 100% accuracy with a less complex dataset. Additionally, there is no explanation of how the algorithms affect the last prediction, and of how one should look at the result if they are not involved in technical matters in the experiment.

A Clinical Decision Support System (CDSS) was created by Yahyaoui et al.⁵⁰ to assist doctors or practitioners in diagnosing diabetes. This study used a number of ML methods, including SVM, RF, and deep CNN, to accomplish this goal. RF yielded the best result in computations with an accuracy of 83.67% compared with DL and SVM having 76.81% and 65.38%.

Naz and Ahuja⁴⁸ used free-source PIMA diabetes datasets with the utilization of ANN, NB, DT, and DL in their study. DL method according to the authors of the study is effective in detecting the onset of diabetes with an accuracy of 98.07% on average. PIMA is one of the most studied and basic datasets, which makes it easy to apply traditional and sophisticated ML algorithms. Thus, it is unsurprising that the PIMA Indian dataset has higher accuracy. Moreover, the article does not talk about the interpretability issues of the model and how the model can work with unbalanced data as well as when there are too many missing values. For example, the generated data in the healthcare industry may be of different types and not necessarily characterized, categorized, and already processed in the identical fashion as the PIMA Indian dataset. While it is important to assess the impartiality of algorithms, absence of bias, reliability, and interpretability in the second task, this article suggests that when developing a CDSS, all the more if working with a multiclass classification dataset where data is missing in large quantity, it is good to assess these characteristics.

Reasons include the presentation by Ashiquzzaman et al.⁵¹ of DL for overfitting handling in diabetes datasets. Using the PIMA Indian dataset, the proposed technique gave an accuracy of 88.41%. The writers report that results were considerably enhanced when dropout strategies were implemented within the model and overfitting issues were duly eliminated. However, if we use the dropout technique too often, we spend an overall elevated amount of time training. It is hard to say if the suggested model for computational time will be the best one, as they did not take these factors into account in their analysis.

Alhassan et al.⁵² presented the King Abdullah International Research Center for Diabetes (KAIMRCD) dataset, the world's largest diabetic dataset with data from 14,000 individuals. In the experiment, the author's CDSS architecture achieved an accuracy of 97% using LSTM- and GRU-based deep NN. Table 4 gives a brief review of studies that used ML and DL techniques for diabetic diagnosis.

ML for diabetes prediction was explored by Febrian et al.,⁵³ where the KNN and Naïve Bayes were compared. They evaluated the confusion matrix for diabetes prediction using health attributes and found that Naïve Bayes significantly beats out KNN. Moreover, Modak and Jha,⁵⁴ developed an innovative diabetes prediction model using several ML techniques, including LR, SVM, Naïve Bayes, and RF. They enhanced the model with ensemble methods such as XGBoost, LightGBM, CatBoost, AdaBoost, and Bagging. Among these, CatBoost achieved the highest accuracy of 95.4%, outperforming XGBoost, and demonstrated a superior AUC-ROC score of 0.99, showcasing its potential for more accurate diabetes forecasting.

The studies on diabetes detection using ML techniques show a variety of models and strong performance across different datasets. The Dirichlet Process Mixture (DPM) achieved an impressive accuracy of 96.74%, while RF reported an AUC of 0.80 on the DIABIM-MUNE dataset.

Table 4 | Comparison of Machine Learning Techniques in Diabetes Detection

Study	Year	Algorithms Utilized	Dataset Used	Data Format	Performance Metrics
55	2019	Dirichlet Process Mixture (DPM)	Proprietary dataset	Tabular	DPM achieved an accuracy of 96.74%.
56	2021	RF	DIABIM-MUNE	Tabular	RF reported an AUC of 0.80.
57	2020	KNN	Proprietary dataset (4900 samples)	Tabular	KNN attained an accuracy of 99.9%.
58	2018	SVM, Decision tree, ANN, logistic Regression	Proprietary dataset	Tabular	SVM demonstrated an accuracy of 79.5% with an AUC of 0.839.
59	2021	PSO, MLPNN	Proprietary dataset	Tabular	PSO achieved an accuracy of 98.73%.
53	2022	KNN, Naïve Bayes	Diabetes dataset	Tabular	Naïve Bayes outperformed KNN in accuracy.
54	2024	Logistic regression, SVM, Naive Bayes, RF, XGBoost, LightGBM, CatBoost, AdaBoost, Bagging	Kaggle Diabetes dataset	Tabular	CatBoost achieved an accuracy of 95.4% and an AUC-ROC of 0.99.

KNN demonstrated high accuracy, reaching 99.9% on a proprietary dataset, while SVM achieved 79.5% accuracy with an AUC of 0.839. The particle-swarm-optimization-based MLP neural network (PSO-MLPNN) model reached an accuracy of 98.73%. Additionally, models like Naïve Bayes outperformed KNN in accuracy, and the more recent study using a range of algorithms on the Kaggle diabetes dataset saw CatBoost achieving 95.4% accuracy and an AUC-ROC of 0.99, showcasing the potential of ensemble methods and advanced models for diabetes detection.

COVID-19

The most significant problem of the century is the highly contagious illness linked to the novel severe acute respiratory syndrome coronavirus 2 (SARS-CoV-2), sometimes referred to as COVID-19. Normally people did not have access to the vaccine throughout the crisis, despite the fact that it was being provided because of the worldwide pandemic.⁶⁰ The high transmissibility and vaccine resistance of the novel COVID-19 Omicron strain are additional causes for concern. The real-time reverse transcription-polymerase chain reaction (RT-PCR) is currently the gold standard for diagnosing COVID-19 infection.⁶¹ The researcher suggested more so that ML and AI can be employed together with more sophisticated tools such as X-ray and CT scan to detect individuals who might be susceptible to the outbreak. For instance, Chen et al.⁶² suggested a UNet++ model with CT images from 51 COVID-19 and 82 non-COVID-19 patients, with their accuracy being 98.5%. Abbasian et al.⁶³ compare the performance of 10 distinct DL models using a tiny dataset of 108 and 86 COVID and non-COVID respectively, achieving an overall accuracy of 99%. It is Wang et al.⁶⁴ who developed an inception-based model using a ton of data and 453 CT scan pictures in order to achieve 73.1% accuracy. Unfortunately, the region of interest (ROI) and network activity of the model were not well defined. The COVNet model was presented by Lin et al.⁶⁵ using a large dataset of 4356 positive chest CT images of pneumonia patients and 1296 positive chest CT images of COVID-19 patients, with an accuracy of 96%.

At the same time, several reports came to recommend using chest X-ray pictures for COVID screening (such as those by Lababidi et al., Khoudour et al., and Narin et al.⁶⁶⁻⁶⁸). Additionally, the other studies made use of larger datasets in order to produce more sophisticated screening methods. For the purposes of the current research, Brunese et al.⁶⁹ used 6505 scans with a 1:1.17 data ratio, of which 3520 were classified as “other patients” and 3003 as COVID-19 symptoms. Ghoshal and Tucker⁷⁰ obtained 92.9% accuracy with a dataset of 5941 images. None of the two studies considered the issue of how the proposed models would perform when presented with data that is skewed and has different class distributions. Apostolopoulos and Mpesiana⁷¹ employed a convolutional-neural-network-based Xception model on a dataset of COVID-19 and non-COVID-19 cases. The dataset included chest X-ray scans of 284 COVID-19 and 967 non-COVID-19 patients, with an average accuracy of 89.6%.

Solayman et al.⁷² developed an ML-based intelligent web application for COVID-19 detection, using techniques such as LR, RF, DT, k-nearest neighbor, SVM, ensemble models (AdaBoost, XGBoost), and DL (artificial neural network, CNN, LSTM). They applied the SMOTE technique for data balancing and the LIME framework for explainable AI, with the hybrid CNN-LSTM model achieving 96.34% accuracy and a 0.98 F1 score on a dataset obtained from the Israeli Ministry of Health. Moreover, Nanekaran et al.⁷³ focused on predicting the environmental impact of pandemic plastics using a DNN model. Their study used data from February 2020 to October 2021 on COVID-19 spread and PPE usage, with the DNN model outperforming other algorithms like KNN, DT, and RFs, achieving an AUC of 0.929 and lower error rates (MSE = 0.024, RMSE = 0.027).

In Table 5 below, the present study identified some of the works that utilized ML and DL techniques to diagnose COVID-19.

The studies on COVID-19 detection using ML techniques highlight the strong performance of CNN across various datasets, with accuracy values ranging from 86% to 98.5%. Notably, CNN models demonstrated accuracy of 94.1% and 95.38% on

Table 5 | Comparison of Machine Learning Techniques in COVID-19 Detection

Study	Year	Algorithms Utilized	Dataset Used	Data Format	Performance Metrics
65	2020	CNN	Mixed dataset	Image	CNN achieved an accuracy of 90%.
62	2020	CNN	Mixed dataset	Image	CNN reported an accuracy of 98.5%.
74	2021	CNN	Mixed dataset	Image	CNN attained an accuracy of 86%.
75	2020	CNN	Cohen's dataset	Image	CNN achieved an accuracy of 94.1%.
76	2020	CNN	Cohen's dataset	Image and Tabular	CNN demonstrated an accuracy of 95.38%.
72	2023	Logistic regression, RF, decision tree, KNN, SVM, AdaBoost, XGBoost, CNN, LSTM	COVID-19 symptoms dataset (Israeli Ministry of Health)	Tabular	Highest accuracy was achieved 96.34%.
73	2023	Deep neural network (DNN), KNN, decision tree, RF, SVM, Gaussian Naïve Bayes, logistic regression, multilayer perceptron	COVID-19 spread and PPE usage data (Iran)	Tabular	Highest accuracy was achieved 0.929.

Cohen's dataset, and 90% on a mixed dataset. Additionally, a combination of models like LR, RF, DT, KNN, SVM, AdaBoost, XGBoost, CNN, and long short-term memory (LSTM) achieved a high accuracy of 96.34% on a COVID-19 symptoms dataset. Another study employing a variety of models on COVID-19 spread and PPE usage data reported an accuracy of 92.9%, demonstrating the effectiveness of DL and traditional ML models for detecting and analyzing COVID-19.

Other Diseases

Both ML and DL have been used to detect diseases other than the ones listed above. There are two key causes for this rising application: big data and the progress in computer hardware. For example, Mao et al used eye movement to classify diseases using DT and RF.⁷⁷ When designing automatic skin disease categorization systems, Nosseir and Shawky⁷⁸ compared KNN with SVM; KNN performed best, with an accuracy of 98.22%. Khan et al.⁷⁹ classified multimodal brain cancers using CNN-based models such as VGG16 and VGG19. Three publicly accessible image datasets were used in the experiment: Specifically, the proposed method achieved an accuracy of 97.8%, 96.9%, and 92.5% when submitted to BraTs2015, BraTs2017, and BraTs2018. Similar work was done by Imran et al.,²⁸ who used the RF classifier for tumor segmentation. Using the datasets BRATS 2012, BRATS 2013, BRATS 2014, BRATS 2015, and ISLES 2015, the authors obtained overall accuracy of 98.7%, 98.7%, 98.4%, 90.2%, and 90.2%, respectively.⁸⁰

Dai et al.⁸¹ developed a CNN for the development of an application for the detection of skin cancer. Using the publicly available HAM10000 dataset, the authors experiment and achieve an accuracy of 75.2%. In 2020, in their work Dai et al.⁸¹ used KNN, SVM, CNN, and majority voting to identify melanoma skin cancer using the ISIC dataset. At 88.4% majority voting achieved the highest accuracy. A list of some of the

literature reviewed in this study using ML and DL in disease diagnosis is presented in Table 6.

The studies on various disease detection using ML techniques show a diverse range of successful applications. For pediatric colonic inflammatory bowel disease, RF achieved 100% accuracy. In liver disease classification, SVM demonstrated accuracy between 90% and 92%. For brain tumor diagnosis, CNN achieved accuracies between 90% and 99%, while RF performed well in brain tumor segmentation with an 88% Dice overlap. In melanoma detection, SVM with feature extraction achieved 96% accuracy, and DT reported 82.35% for skin cancer detection. Thermal imaging for skin cancer showed a precision of 0.9665 with EL. In hepatocellular carcinoma (HCC) detection, InceptionV3 reported accuracy between 89% and 96%, while RF models demonstrated an AUC of 0.803 for predicting HCC postoperative death outcomes, indicating the wide applicability and success of ML techniques in medical diagnostics.

Discussion

A significant reliance on ML, EL, and DL models is found in the literature to perform disease classification across diverse medical datasets as shown in Figure 2. These ML models (RF and SVM) are employed as they have proved to deliver robust performance on image as well as tabular datasets, as evident in studies of Dhaliwal et al.,⁸² Waheed et al.,⁸⁶ and Wang et al.,⁹⁰ which showed that EL techniques like gradient boosting and AdaBoost can combine many weak learners into a more reliable and accurate predictor. For image-based diagnosis tasks like brain tumor detection⁸⁴ and melanoma classification,⁸⁸ CNNs are often used as DL models to extract the best characteristics from complex data. The reason these methods are widely adopted is that they have been shown to enhance diagnostic accuracy and to cope with different data formats and complexities.

It also describes how the choice of algorithm is strategically constrained by disease dynamics and

Table 6 | Comparison of Machine Learning Techniques in Various Disease Detection

Study	Disease	Algorithms Utilized	Dataset Used	Data Format	Performance Metrics
82	Classification of pediatric colonic inflammatory bowel disease	RF	74 proprietary cases	Image	RF achieved an accuracy of 100%.
83	Liver disease classification	SVM	ILPD, BUPA	Tabular	SVM reported an accuracy of 90%–92%.
84	Brain tumor diagnosis	CNN	Brain tumor challenge sites and MRI	Image	CNN demonstrated an accuracy range of 90%–99%.
85	Brain tumor segmentation	RF	MICCAI, BraTS 2013	Image	RF achieved an 88% Dice overlap.
86	Melanoma detection using dermoscopic images	SVM with color and feature extraction	PH2	Image	SVM attained an accuracy of 96%.
87	Melanoma skin cancer detection	Naïve Bayes, decision tree, KNN	MED-NODE	Image	A decision tree classifier reported an accuracy of 82.35%.
88	Skin cancer detection with thermal imaging	EL and DL	CSLTAD	Image	EL achieved a precision of 0.9665.
89	Hepatocellular carcinoma detection	InceptionV3	Genomic Data Commons	Image	InceptionV3 reported an accuracy of 89%–96%.
90	Prediction of HCC postoperative death outcomes	RF, gradient boosting, GBM, logistic regression, decision tree	BioStudies database	Tabular	A random forest model demonstrated an AUC of 0.803.

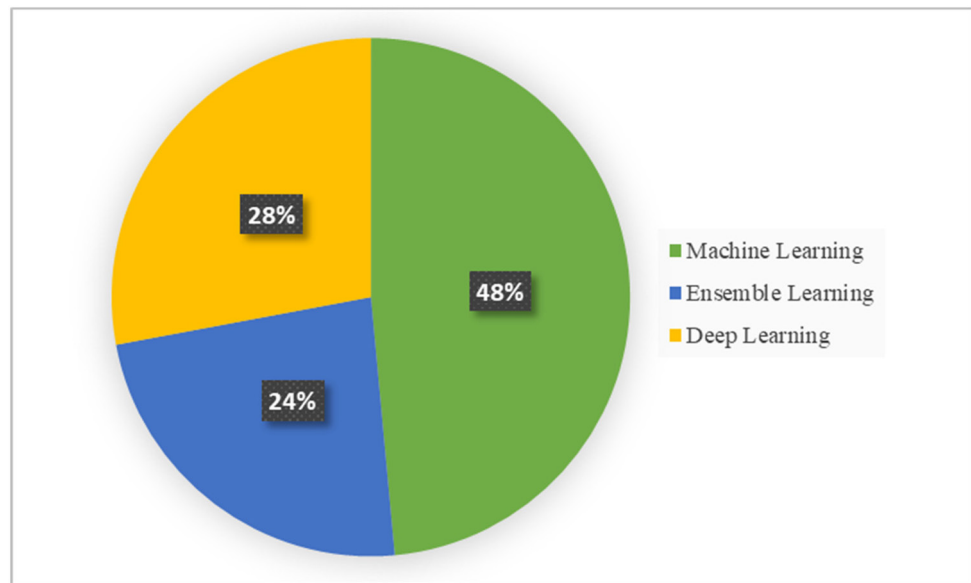


Fig 2 | Distribution of models among the studies

data properties. Due to excellent high accuracy, ML models are dominating breast cancer detection⁴¹ and heart disease classification¹³ owing to their capability to work well with structured tabular data. For the medical prediction of diseases like diabetes,⁵⁴ and liver cancer,⁹⁰ one of the EL techniques, gradient boosting and RF are often used due to the reliability in prediction by combining multiple models. Because CNNs, a subset of DL models, are capable of obtaining important characteristics from high-dimensional image data, they are chosen for imagery-focused tasks like COVID-19 detection⁷² and brain tumor diagnosis.⁸⁴

The disease-specific requirements and the strengths of each algorithm type were aligned when selecting models to suit the diversity of diagnostic applications without diminishing performance. The mapping of diseases and models is shown in Figure 3.

Computational models evolved over the years and are increasingly being adopted and improved for medical diagnosis and prediction tasks as shown in Figure 4. From 2015 to 2016 though, the simplicity and effectiveness of tabular and image data made the landscape dominated by ML models like LR, SVM,⁴³ etc. In 2017 and 2018, there has been a rapid increase

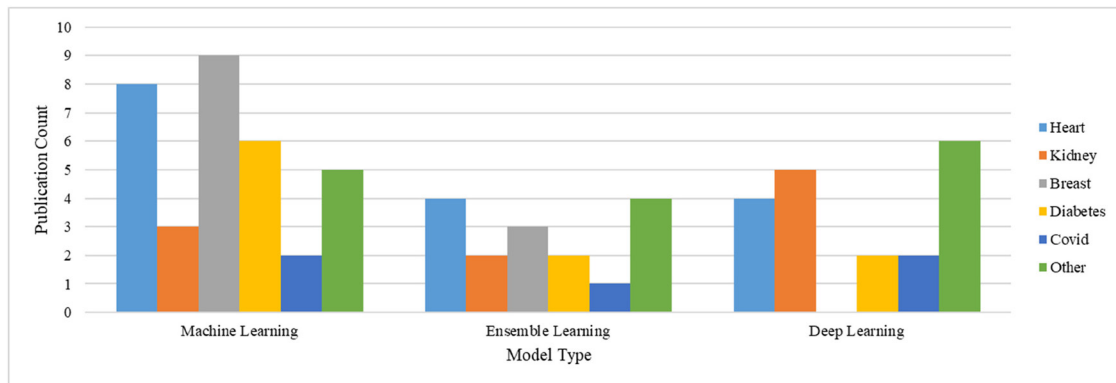


Fig 3 | Disease and model mapping

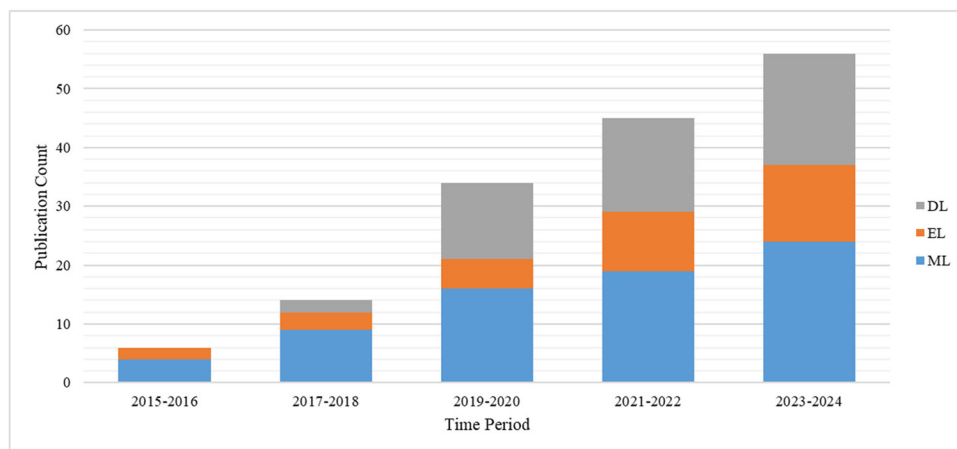


Fig 4 | Evolution of models in early disease detection

in the usage of DL models (e.g., CNNs),⁴⁵ thanks to the increasing availability of computational power and medical imaging data. Due to this, by 2019–2020, ensemble learning (EL) techniques, which include RF and gradient boosting,⁵⁷ became popular aligners with higher prediction accuracy through model aggregation; and DL models for fast integration of tasks that need complex feature extraction in the diagnosis of brain tumor.⁸⁴ The model types for all three models exponentially grew between 2021 and 2022, aided by more diverse data and hybrid approaches.⁷² The period from 2023 to 2024 has seen a significant increase in the adoption of deep learning (DL) and ensemble learning (EL) methods. For example, research has highlighted the effectiveness of the CatBoost algorithm in predicting diabetes, and a hybrid model combining Convolutional Neural Networks (CNN) and Long Short-Term Memory (LSTM) networks has shown promise in detecting COVID-19; this explains the move from much easier and less moil category of methods for medical analytics to more advanced and data exclusive.

Challenges and Opportunities in Early Disease Detection

A tide of potential healthcare advancement comes with incorporating ML into early disease detection. However, it is also a challenging environment that

goes beyond technical complexities. When embarking on the exciting path of leveraging predictive power for detecting diseases at an early stage, we must acknowledge the ethical and technical foundations on which this transformation rests. Sensitivities in the healthcare industry revolve around personal and medical data, which constitute highly sensitive medical information. Secure data storage, transmission, and access controls are necessary to protect patient privacy. A long-standing challenge is to strike an equilibrium between the use of data for early detection models and the protection of patient privacy.⁹¹

Before using patient data in predictive models, we must have consent. A key aspect of establishing trust between patients and healthcare providers involves transparent communication of what data might be used for, potential hazards associated as well as potential benefits. When training models of ML on historical data, any inherent biases in that data can be unconsciously learned by those models. This can lead to unfair predictions that have been shown to disproportionately affect members of certain demographic groups. Eliminating bias and achieving fairness in predictive models are critical for achieving equitable healthcare outcomes. Since ML models tend to be increasingly elaborate, they have a tendency to turn into something like “black

boxes,” and it is often hard to understand how exactly they come to make these predictions. If models are not explicable and comprehensible⁹² then healthcare professionals will not be able to rely on and act upon their predictions.

The integration of ML into healthcare workflows might remain a cause for concern in the case of inaccurate predictions and whether an entity should be accountable for if and when incorrect predictions are made. It is difficult to determine responsibility and set up rules for corrective actions in these situations. How representative and good the training data is, determines how precise the predictive models will be. In addition, erroneous or insufficient data often leads to inaccurate predictions. Therefore, thorough data preprocessing and duration for getting the prediction are required. However, to validate that predictive models are dependable and generalizable, they must be validated on diverse and independent datasets. A technical issue to be solved is the problem of overfitting, which occurs when a model trains well on the training data, but poorly on new data.

Certain ML methods are too complicated to use in medical environments. In order to build trust, models need to be developed that generate interpretable insights so that healthcare professionals can understand the reasoning behind predictions. Integrating predictive models into current clinical workflows is a technical challenge. To be integrated seamlessly, it has to be compatible with electronic health records, imaging systems, and other healthcare technologies. Since healthcare data is becoming larger and larger, predictive models need to be scalable to process a large dataset efficiently. Models are still effective as the volume of data increases, due to scalability. This is a technical challenge when real-time predictions are needed and predictive models need to rapidly process data and are able to produce practical insights in a timely fashion. Multiple stakeholders need to collaborate to steer the intersection of ethical and technical considerations in the predictive detection of diseases.⁹³ Only multidisciplinary teams including healthcare professionals working with data scientists, ethicists, legal experts, and policymakers can design ethical guidelines and structures. ML holds enormous potential to revolutionize the healthcare industry through predictive disease detection. Yet the voyage is beset with ethical and technical hurdles that have to be surmounted in order for a responsible, equitable implementation. Not only is it necessary for successful adoption, but it is also necessary to strike the right balance between improving technology and keeping ethical values in order to get the maximum benefits from predictive power and the minimum associated risks (Table 7).

A Glimpse into the Future: Emerging Trends and Prospects in Machine-Learning-Driven Early Disease Diagnostics

The application of ML to early disease detection has led to significant healthcare advances, but the road ahead is promising. As technology and our understanding of

diseases advance, ML-based early disease detection appears to be on the brink of an emerging trend that could shape the future of healthcare. These trends have the potential to revitalize the healthcare system while improving our capacity to identify diseases early on. Predictive model optimization will require the integration of data from multiple sources, such as wearable technologies, medical imaging, genomic data, and electronic health records. A more accurate and detailed picture of a patient's health status will be provided by ML algorithms that integrate many data modalities, such as test results, radiological images, prescription drugs, and vital signs. The need for model interpretability is growing as ML models get more intricate. The rise of explainable AI, which seeks to create algorithms that can provide an explanation for their predictions, allays this worry. Gaining the trust of medical experts and empowering them to base their decisions on the algorithmically offered insights will depend on this. Transfer learning is the process of training ML models for some conditions and then using what they have learned to identify other diseases that are similar. The versatility of this approach enables models created for one disease to be altered for the early diagnosis of related diseases, boosting output and speeding up model development.⁹⁴

There is an abundance of gadgets, and ongoing health monitoring, as a result of which ML algorithms can now predict and issue real-time alerts. This also benefits both patients and medical professionals by giving them timely notification about upcoming health issues so prompt treatment and prevention can take place. Although quantum computing is still in its infancy, the impact on the healthcare sector will be huge. The consequent processing power of this could help the training of and tinkering around with complex ML models, ultimately speeding and refining the process of disease diagnosis. ML is capable of analyzing large datasets and can help to build precision medical strategies. Predictive models combine panoply into an individual's genetic fingerprint, lifestyle, and medical history to inform the development of personalized treatment plans.⁹⁵

Applications of LLMs in Early Disease Detection

The integration of large language models (LLMs) in early disease detection offers significant promise, particularly in handling unstructured healthcare data such as clinical notes, medical literature, and patient records. LLMs can process and analyze vast amounts of text data to identify patterns, correlations, and risk factors that might otherwise go unnoticed.⁹⁶ By leveraging their ability to understand context and draw inferences from natural language, LLMs can assist healthcare professionals in diagnosing diseases at earlier stages by providing insights into a patient's medical history, symptoms, and potential risk factors. Additionally, LLMs can enhance decision support systems by synthesizing research findings and clinical guidelines, helping doctors make more informed, data-driven decisions.⁹⁷

Furthermore, LLMs can be instrumental in analyzing and predicting trends in disease outbreaks or patient

Aspect	Challenges	Solutions/Considerations
Data Privacy and Security	Sensitive personal and medical data require secure storage, transmission, and access controls to protect patient privacy.	Ensure consent before using patient data; Implement strong security measures to protect data; Develop transparent communication strategies with patients about data usage and potential risks/benefits.
Bias in Data	Machine learning models may inherit biases from historical data, leading to unfair predictions and disproportionately affecting certain demographic groups.	Eliminate biases and ensure fairness in models; Regularly audit models for discriminatory outcomes.
Model Explainability	Complex models may act as “black boxes,” making it hard for healthcare professionals to understand how predictions are made, affecting trust and reliability.	Develop interpretable models that provide clear explanations for predictions; Ensure healthcare professionals can act on the model’s insights.
Accountability for Inaccurate Predictions	Uncertainty about who is responsible for incorrect predictions and the corrective actions needed.	Establish clear accountability rules; Define corrective actions for incorrect predictions; Ensure reliable and accurate data is used for model training.
Data Quality and Preprocessing	Erroneous or insufficient data can lead to inaccurate predictions, making data preprocessing and model validation essential.	Ensure high-quality, diverse data is used for training; Perform thorough data preprocessing; Validate models using independent datasets to improve accuracy and generalizability.
Overfitting	Overfitting occurs when models perform well on training data but poorly on new, unseen data.	Regularize models, use cross-validation techniques, and ensure diverse datasets to prevent overfitting.
Model Complexity and Usability	Some machine learning methods are too complex for use in medical environments, affecting adoption and trust.	Develop simpler, interpretable models that generate understandable insights for healthcare professionals.
Integration into Clinical Workflows	Integrating predictive models into existing healthcare technologies like electronic health records and imaging systems is a technical challenge.	Ensure models are compatible with current systems; Work with healthcare professionals to streamline integration into daily clinical workflows.
Scalability	Predictive models need to efficiently process large datasets, particularly as healthcare data continues to grow.	Design scalable models that can handle increasing volumes of data without compromising speed or accuracy.
Real-Time Predictions	Real-time predictions require models that can process data quickly and deliver practical insights in a timely manner.	Optimize models for real-time prediction and response, ensuring low latency for time-sensitive decision-making.
Multidisciplinary Collaboration	Developing ethical and technical guidelines requires input from multiple stakeholders, including healthcare professionals, data scientists, ethicists, legal experts, and policymakers.	Encourage collaboration across disciplines to create comprehensive ethical frameworks and technical solutions for responsible predictive disease detection.

health conditions. Their ability to continuously learn from diverse datasets, including patient reports and scientific publications, enables them to track emerging patterns in disease progression or responses to treatment.⁹⁸ For example, LLMs can assist in identifying early warning signs for conditions like cancer, cardiovascular diseases, or neurological disorders by analyzing subtle language cues from patient interactions and medical histories. Their adaptability allows for real-time updates on disease trends, which could lead to more timely interventions. Ultimately, the application of LLMs in early disease detection not only augments the capabilities of medical professionals but also represents a transformative shift towards more proactive, predictive, and personalized healthcare.

Conclusion

This study underscores the significant impact of ML, EL, and DL models in early disease detection, with ML models being the most widely applied across various healthcare domains. The findings highlight the evolution of these models from simpler ML techniques to more sophisticated DL and ensemble approaches, demonstrating the increasing capacity to handle complex medical data and enhance

diagnostic precision. However, challenges remain, particularly in areas such as data privacy, model interpretability, and the integration of these technologies into clinical workflows. Despite these obstacles, the study emphasizes how emerging trends and continuous advancements in AI-driven models hold the transformative potential to revolutionize early disease detection. This could lead to more proactive and personalized healthcare, ultimately improving patient outcomes, reducing healthcare costs, and advancing healthcare equity by providing better access to accurate diagnostic tools. As these technologies evolve, they will not only benefit the medical community but also have a profound societal impact by ensuring that earlier, more accurate diagnoses contribute to better overall health outcomes for individuals and communities.

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