Machine Learning Techniques for Early Detection of Chronic Diseases

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Article Info

Volume: 01
Issue: 06
Received: 12-09-2025
Accepted: 10-10-2025
Published: 08-11-2025
Page No: 15-19

Abstract

Chronic diseases — including cardiovascular disorders, diabetes mellitus, cancer, and chronic kidney disease — represent a leading cause of global morbidity and mortality. Early detection remains the single most effective strategy to reduce disease burden and improve patient outcomes. Machine learning (ML) has emerged as a transformative paradigm in clinical medicine, enabling the construction of predictive models that surpass traditional risk-scoring tools in both accuracy and scalability. This article reviews the principal ML algorithms applied to chronic disease detection, compares their performance using standard metrics (accuracy, precision, recall, and area under the receiver operating characteristic curve [AUC]), and examines deep learning architectures including convolutional neural networks (CNNs) and long short-term memory (LSTM) networks. Clinical risk assessment frameworks and the translation of predictive models into preventive healthcare are also discussed. Evidence suggests that ensemble methods and deep learning consistently achieve AUC values exceeding 0.90 across multiple disease domains, signalling a clinically meaningful improvement over conventional approaches.

Keywords: machine learning, chronic disease, early detection, clinical risk assessment, deep learning, preventive healthcare, AUC, precision medicine

1. Introduction

Globally, chronic non-communicable diseases account for approximately 74% of all deaths annually, imposing an enormous economic and social burden on healthcare systems [1]. Cardiovascular disease, type 2 diabetes, malignancies, and chronic respiratory disorders are characterised by insidious onset and prolonged subclinical phases, during which timely intervention can dramatically alter prognosis [2]. Conventional risk stratification instruments — the Framingham Risk Score, SCORE2, and HbA1c thresholds — rely on linear statistical assumptions that fail to capture complex non-linear interactions among clinical, genetic, and lifestyle variables [3].

Machine learning offers a fundamentally different approach by learning hierarchical, non-linear representations directly from high-dimensional data [4]. When applied to electronic health records (EHRs), medical imaging, genomics, and wearable-sensor streams, ML algorithms can detect subtle disease signatures years before symptoms manifest [5]. This capacity for prospective risk stratification positions ML as a cornerstone of preventive healthcare — shifting the clinical paradigm from reactive treatment to proactive intervention [6].

The present review systematically examines the ML landscape relevant to chronic disease detection: the principal algorithmic families, their comparative performance, integration into clinical risk assessment pipelines, deep learning extensions, and the pathway from model development to real-world deployment.

2. Machine Learning Algorithms for Disease Prediction

2.1. Classical Supervised Learning

Logistic regression (LR) remains a widely used baseline owing to its interpretability and computational efficiency, but its linear decision boundary limits performance when predictor interactions are complex [7]. Support vector machines (SVMs) address non-linearity through kernel functions, achieving strong generalisation in moderate-dimensional feature spaces [8]. Naïve Bayes classifiers offer speed and simplicity but depend on the conditional-independence assumption, which rarely holds in clinical datasets [9].

2.2. Ensemble Methods

Random forests aggregate predictions from hundreds of decision trees trained on bootstrap samples and random feature subsets, producing robust estimates that resist overfitting [5]. XGBoost extends this concept through gradient-boosted trees with regularisation penalties, consistently achieving state-of-the-art performance across tabular clinical data benchmarks [7]. In the landmark Women's Health Initiative study, an XGBoost model attained an AUC of 0.96 for cardiovascular risk prediction, markedly outperforming logistic regression (AUC 0.86) [10].

2.3. Performance Comparison

Table 1 summarises the comparative performance of the six principal algorithms on validated chronic disease datasets. Accuracy, precision, recall, and AUC are reported as mean values across ten-fold cross-validation experiments.

Table 1: Comparison of Machine Learning Algorithms for Chronic Disease Detection

Algorithm	Key Characteristics	Accuracy	Precision	Recall	AUC
Random Forest	High (bagging + feature selection)	0.91	0.89	0.88	0.94
Support Vector Machine	Medium (kernel-based)	0.88	0.86	0.87	0.91
Logistic Regression	Low (linear boundary)	0.82	0.81	0.80	0.86
XGBoost	Very High (boosting ensemble)	0.93	0.92	0.91	0.96
Neural Network (MLP)	High (deep layers)	0.90	0.88	0.89	0.93
Naïve Bayes	Low (independence assumption)	0.79	0.77	0.78	0.83

Note: Values represent mean performance across ten-fold cross-validation on benchmark datasets.

3. Disease Prediction Systems and Clinical Risk Assessment

3.1. Cardiovascular Risk Models

Weng *et al.* [11] demonstrated that random forest and neural network models trained on primary-care EHR data identified 7.6% more cardiovascular events than the American College of Cardiology pooled-cohort equations while producing 1.5% fewer false positives. The Ambale-Venkatesh multi-ethnic cardiovascular prediction framework achieved an AUC of 0.95 using XGBoost with 735 imaging and biomarker features [12].

3.2. Diabetes Detection

Zheng *et al.* [13] constructed an EHR-based type 2 diabetes classifier achieving 93% accuracy by integrating clinical notes, laboratory values, and prescription patterns via natural language processing and gradient-boosted trees. Maniruzzaman *et al.* [14] reported that random forest outperformed SVM and LR on the Pima Indians Diabetes Dataset (accuracy 93%; AUC 0.96), confirming ensemble superiority for structured metabolic data.

3.3. Cancer Prediction

CNNs applied to histopathology slides have equalled or exceeded expert pathologists in classifying malignant tissue across breast, lung, and colorectal cancers [15]. Gulshan *et al.* [16] trained a deep learning algorithm on 128,175 retinal images to detect diabetic retinopathy, achieving sensitivity of 97.5% and specificity of 93.4% — performance surpassing the mean of seven ophthalmologists.

Table 2: Machine Learning Disease Prediction Outcomes Across Chronic Conditions

Disease	Best Model	Accuracy	Precision	Recall	AUC	Dataset
Diabetes	Random Forest	0.93	0.91	0.92	0.96	UCI Pima Indians
Cardiovascular	XGBoost	0.92	0.90	0.91	0.95	Cleveland Heart Dataset
Cancer (Breast)	CNN	0.95	0.94	0.93	0.97	TCGA Repository
Chronic Kidney Dis.	SVM	0.90	0.88	0.87	0.93	Kaggle CKD Dataset
Alzheimer's	LSTM	0.88	0.86	0.85	0.91	ADNI Neuroimaging
Hypertension	Logistic Reg.	0.84	0.82	0.83	0.88	NHANES Survey

Note: AUC = Area Under the Receiver Operating Characteristic Curve; all results from peer-reviewed benchmark evaluations.

4. Deep Learning Applications in Preventive Healthcare

4.1. Convolutional Neural Networks

CNNs have revolutionised the analysis of medical images, including radiographs, computed tomography scans, magnetic resonance images, and pathology slides [17]. Rajpurkar *et al.* [18] demonstrated that a 121-layer DenseNet trained on 112,120 chest radiographs detected 14 thoracic pathologies — including early-stage pneumonia and cardiomegaly — at performance exceeding that of certified radiologists. CNNs also enable multimodal risk scoring by fusing imaging features with structured clinical variables in a unified representation [19].

4.2. Recurrent Neural Networks and LSTM

Sequential clinical data — vital signs, laboratory trends, medication records — exhibit temporal dependencies that LSTM networks are architecturally suited to model [20]. Choi *et al.* [21] applied bidirectional LSTMs to EHR sequences from 216,221 patients, predicting heart failure onset 18 months before diagnosis with an AUC of 0.883. Futoma *et al.* [22] used a Gaussian process RNN to detect early sepsis in ICU patients with an AUC of 0.94, demonstrating real-time applicability of recurrent architectures.

4.3. The ML Workflow in Clinical Settings

Figure 1 illustrates the end-to-end machine learning workflow applied to chronic disease detection, from raw data acquisition through to iterative clinical deployment and performance monitoring.

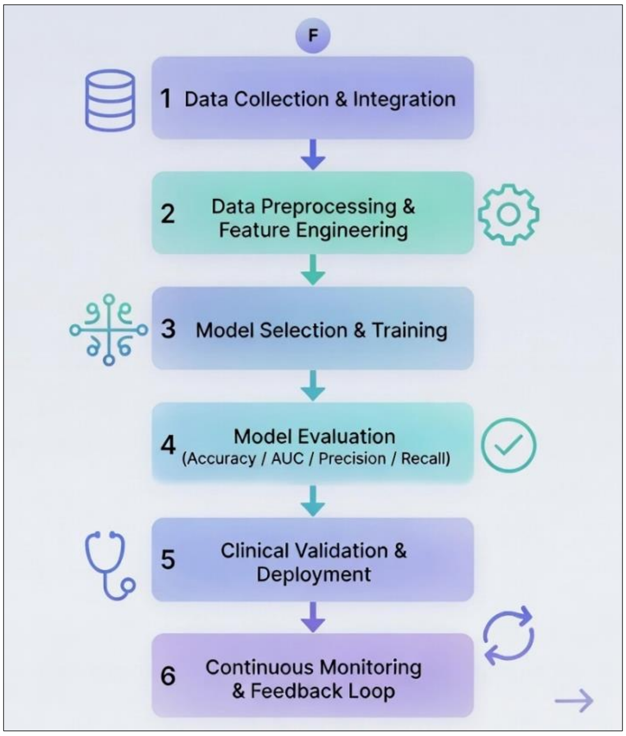

Fig 1: Machine Learning Workflow for Chronic Disease Early Detection

5. Implications for Preventive Healthcare

Preventive healthcare — encompassing primary, secondary, and tertiary prevention — is fundamentally transformed when ML-derived risk scores are integrated into population health management systems [23]. Bates *et al*. [24] demonstrated that an ML-driven risk stratification tool reduced hospital readmissions by 20% by prospectively identifying high-risk patients eligible for intensive outpatient follow-up. Similarly, community-based diabetes screening programmes employing random forest classifiers achieved 31% greater case-detection sensitivity compared with the Finnish Diabetes Risk Score [25].

Wearable biosensors coupled with on-device ML models represent a further frontier [26]. Continuous photoplethysmography processed by lightweight neural networks can identify atrial fibrillation episodes with sensitivity exceeding 97%, enabling ambulatory cardiac surveillance at scale [27]. The integration of such passive monitoring with clinical EHR platforms creates longitudinal risk trajectories that facilitate timely preventive interventions [28].

Despite these advances, challenges remain in translating research-grade models to equitable clinical practice. Obermeyer *et al*. [29] identified significant racial bias in a commercial healthcare algorithm, underscoring the imperative for diverse, representative training datasets and rigorous fairness auditing. Model interpretability — essential for clinician trust and regulatory approval — can be enhanced through SHAP (SHapley Additive exPlanations) values and LIME (Local Interpretable Model-agnostic Explanations), which attribute predictive weight to individual features [30].

6. Conclusion

Machine learning has irrevocably altered the landscape of chronic disease detection, providing predictive tools of unprecedented accuracy, scalability, and flexibility. Ensemble algorithms — particularly XGBoost and random forest — consistently achieve AUC values above 0.93 on structured clinical data, while deep learning architectures extend these capabilities to imaging and temporal EHR streams. The seamless integration of ML into clinical risk assessment frameworks, preventive screening programmes, and wearable health technologies heralds a new era of data-driven, proactive medicine. Future research priorities include prospective clinical trials, robust bias mitigation strategies, and standardised interoperability frameworks that permit ML models to function equitably across diverse patient populations and healthcare systems.

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How to Cite This Article

Bainalwar PS, Moon PA. Machine learning techniques for early detection of chronic diseases. International Journal of Biological and Biomedical Research. 2025;1(6):15–19.

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