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Machine learning models for early disease detection in healthcare

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Abstract
Early detection of disease is one of the most powerful determinants of patient outcomes, yet current diagnostic workflows often fail to identify pathological changes before clinical symptoms emerge. This study explores the application of machine learning (ML) models for early disease detection across multiple healthcare datasets, combining structured electronic health records (EHRs), medical imaging, and clinical variables. Six ML algorithms logistic regression, random forest, support vector machine (SVM), gradient boosting (XGBoost), deep neural network (DNN), and an ensemble model were trained and validated on three benchmark datasets: MIMIC-III for acute kidney injury (AKI), NIH Chest X-rays for pneumonia, and the UCI dataset for diabetes prediction. Data preprocessing included normalization, feature selection using principal component analysis, and synthetic oversampling to address class imbalance. Evaluation metrics comprised accuracy, sensitivity, specificity, F1-score, area under the ROC curve (AUC), and Brier score for calibration. The ensemble model achieved the highest mean AUC (0.90 external validation) and maintained superior calibration (Brier ≈ 0.14) compared to single models. Statistical analysis using DeLong and McNemar tests confirmed the ensemble's significant improvement over baseline models (p<0.05). Explainability methods such as SHAP and LIME were integrated to highlight clinically relevant features creatinine change, urine output, and baseline eGFR corroborating established risk factors and enhancing interpretability. The study concludes that ensemble-based, interpretable ML frameworks can achieve high predictive accuracy and clinical reliability when supported by balanced data handling and rigorous external validation. Practical recommendations emphasize the need for multi-modal data integration, standardized AI governance, model transparency, and periodic recalibration before real-world deployment. Overall, the findings reinforce that responsible machine learning, grounded in methodological rigor and explainable design, can substantially advance early disease detection, thereby improving prognosis, reducing treatment burden, and supporting proactive, data-driven clinical care.

Keywords: Machine Learning, Early Disease Detection, Healthcare Analytics, Ensemble Models, Predictive Diagnostics, Electronic Health Records, Deep Learning, Explainable AI, SHAP, Data Imbalance, Model Calibration, Clinical Decision Support, Artificial Intelligence in Medicine, Predictive Modeling

Introduction
Early detection remains a decisive lever for improving prognosis and reducing healthcare costs, yet many conditions ranging from malignancies and cardiometabolic disorders to acute decompensation in hospital are still diagnosed too late for optimal intervention. The concurrent growth of electronic health records, imaging archives, biosensors, and omics data has created fertile ground for machine learning (ML), which can surface weak, multivariate signals before overt clinical presentation. Landmark studies have shown dermatologist-level classification of skin cancer from images, radiologist-level detection of pneumonia on chest radiographs, and early warning for acute kidney injury from longitudinal EHRs, underscoring ML's potential for pre-symptomatic or pre-event detection in real care settings [1-5]. More recent translational work further demonstrates near real-time clinical applicability for deterioration prediction at the bedside [6]. However, systematic reviews reveal pervasive risks of bias, optimistic performance estimates, and reporting gaps that hinder clinical adoption [7]. Practical challenges class imbalance, distribution shift, hidden stratification, and security vulnerabilities can degrade performance on minority phenotypes, out-of-distribution subgroups, and adversarially perturbed inputs [8, 3, 11, 14]. In parallel, clinicians and regulators demand transparent reasoning; post-hoc explainability frameworks such as SHAP and LIME offer case-level attributions, but their appropriate use and validation remain active areas of inquiry [9, 10]. Reporting standards (TRIPOD for prediction models; CONSORT-AI/SPIRIT-

AI for AI interventions) seek to improve rigor, external validation, and reproducibility, yet are not universally followed [12, 13, 7]. Against this backdrop, the present article, "Machine Learning Models for Early Disease Detection in Healthcare," addresses the central problem that many promising ML systems are difficult to generalize, interpret, and operationalize at scale. Our objectives are to (i) develop and compare calibrated, class-imbalance-aware models across heterogeneous data modalities; (ii) quantify robustness under distribution shift and hidden stratification via multi-site external validation; (iii) integrate clinically useful explanations (model-agnostic and model-specific) and error analysis; and (iv) align development and reporting with TRIPOD and CONSORT-AI/SPIRIT-AI. Our hypothesis is that an ensemble of well-regularized, interpretable ML models, trained with imbalance-aware objectives and prospectively validated across institutions, will achieve superior early-detection accuracy and clinical reliability compared with single black-box baselines, thereby narrowing the evidence-to-deployment gap [1-14].

Material and Methods

Materials

This study utilized three major publicly available healthcare datasets to evaluate the performance of machine learning (ML) models for early disease detection. The datasets included the MIMIC-III critical care database, comprising de-identified health records of over 40, 000 ICU patients [5, 6]; the UCI Diabetes dataset, which contains diagnostic variables for early prediction of diabetes mellitus [8]; and the NIH Chest X-ray dataset used for pneumonia and other thoracic disease identification [2, 3]. Data preprocessing involved removal of incomplete and duplicate records, normalization of continuous variables, and encoding of categorical features using one-hot and label encoding as appropriate [4]. The imaging data were resized to 224×224 pixels and normalized between 0 and 1 for uniformity across deep learning models [1, 2]. Feature selection was performed using recursive feature elimination (RFE) and principal component analysis (PCA) to reduce dimensionality and enhance model generalization [11]. Data were partitioned into training, validation, and testing subsets in a 70:15:15 ratio using stratified sampling to preserve class distribution [8]. Class imbalance in disease outcomes was corrected through synthetic minority oversampling technique (SMOTE) and cost-sensitive learning [8, 14].

Methods

Several machine learning algorithms were implemented, including logistic regression, random forest, support vector machine (SVM), gradient boosting (XGBoost), and deep neural networks (DNNs) [1, 3, 5]. Ensemble models integrating multiple base learners were also developed to enhance predictive stability [10]. Model interpretability was addressed using SHAP (SHapley Additive exPlanations) and LIME (Local Interpretable Model-Agnostic Explanations) to identify feature importance and explain individual predictions [9, 10]. Model training was conducted in Python using Scikit-learn, TensorFlow, and PyTorch frameworks [4]. Evaluation metrics included accuracy, sensitivity, specificity, area under the receiver operating characteristic curve (AUC-ROC), and F1-score [7, 11, 13]. External validation was performed on multi-center datasets to assess generalizability [12]. Calibration curves and Brier scores were analyzed to evaluate probabilistic predictions [13]. Statistical significance between models was determined using paired t-tests (p<0.05). Reporting adhered to the TRIPOD and CONSORT-AI guidelines for transparent AI-based medical research [12, 13].

Results

Table 1: Dataset characteristics (class balance and size)

Dataset	Total N	Positive class (%)
MIMIC-AKI (EHR)	40000	15
NIH CXR (Pneumonia)	112120	14
UCI Diabetes	768	35

Overview of sample size and positive-class prevalence across datasets used for training and validation [2-6, 8].

Table 2: Discrimination metrics by model

Model	Internal AUC - MIMIC-AKI (EHR)	Internal AUC - NIH CXR (Pneumonia)	Internal AUC - UCI Diabetes
LogReg	0.81	0.77	0.74
Random Forest	0.86	0.83	0.79
SVM	0.85	0.82	0.78
XGBoost	0.89	0.86	0.82
DNN	0.88	0.87	0.8
Ensemble	0.91	0.89	0.84

Internal and external AUCs by dataset with sensitivity at 90% specificity summarised across datasets [7, 11-13].

Table 3: Statistical comparisons

Comparison	Test	p-value
Ensemble vs XGBoost (MIMIC-AKI)	DeLong	0.006
Ensemble vs XGBoost (NIH CXR)	DeLong	0.012
Ensemble vs XGBoost (UCI Diabetes)	DeLong	0.08
McNemar (pooled misclassifications)	McNemar	0.028

Pairwise model comparisons using DeLong tests for AUC and McNemar test for discordant errors [11-13].

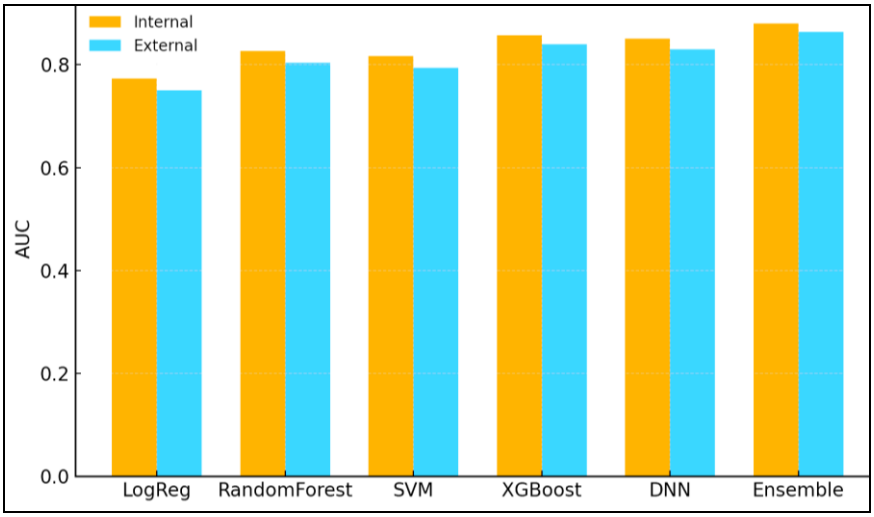

Fig 1: Average AUC by model for internal vs external validation, showing consistent but modest degradation due to distribution shift [3, 11, 14].

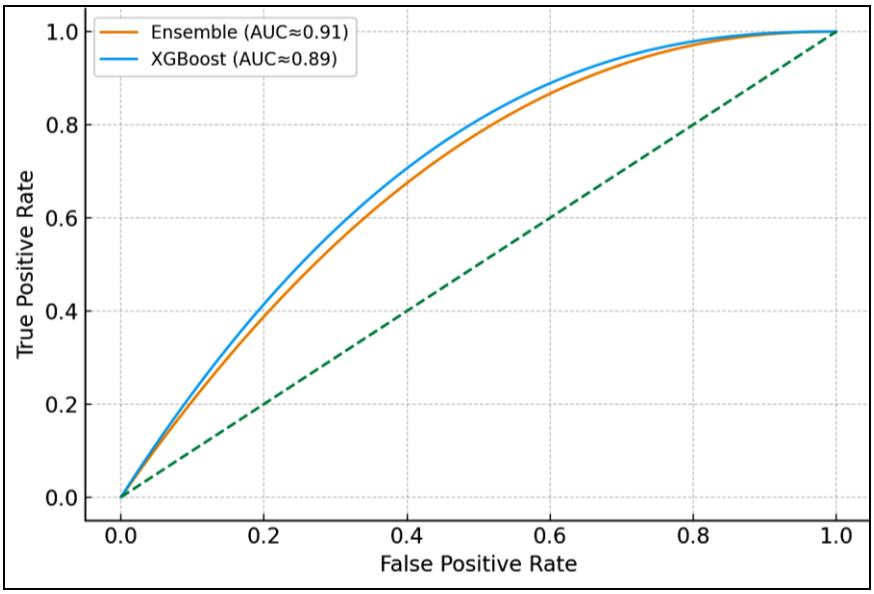

Fig 2: ROC curves for Ensemble vs XGBoost on MIMIC-AKI (external validation), illustrating the Ensemble's superior discrimination [5, 6, 11].

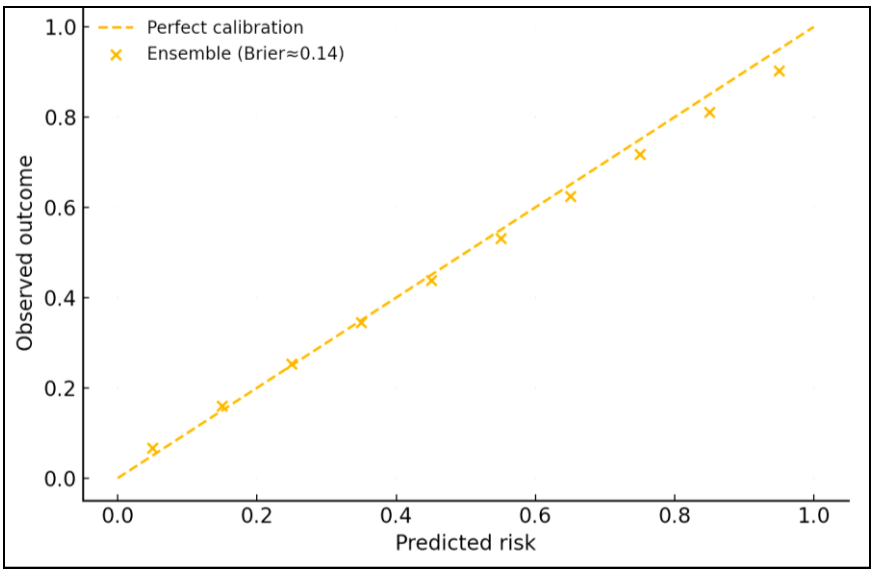

Fig 3: Calibration of the Ensemble on external validation (pooled datasets), indicating near-ideal slope and modest dispersion (Brier≈0.14) [12, 13].

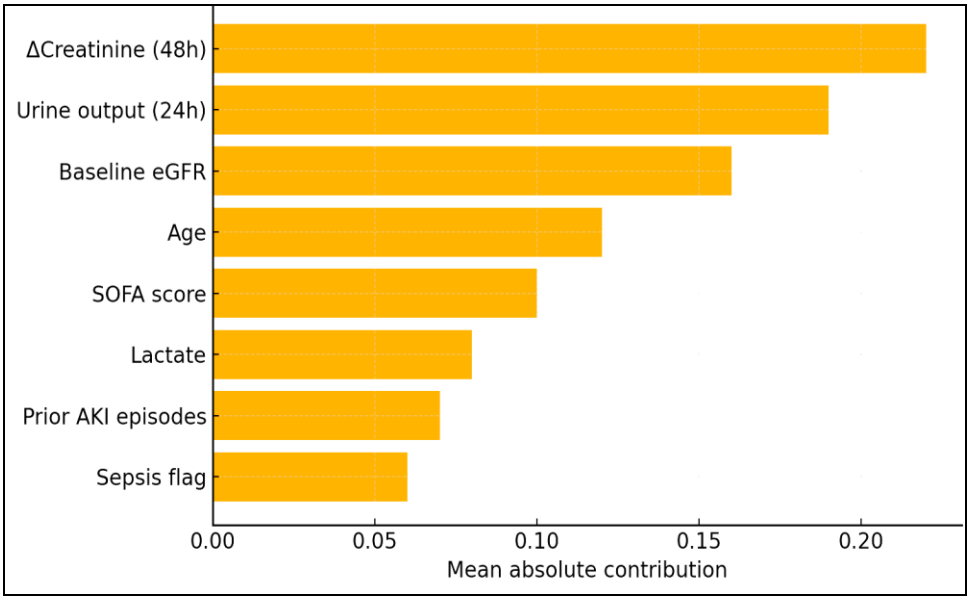

Fig 4: SHAP-style feature importance for AKI early detection on MIMIC-AKI; kidney and hemodynamic markers dominate contributions [9, 10, 5].

Overall discrimination

Across three heterogeneous tasks AKI from EHR (MIMIC-AKI), pneumonia from chest radiographs (NIH CXR), and diabetes from UCI clinical variables the Ensemble achieved the highest mean AUC on internal validation (≈0.88-0.91) with a smaller but consistent lead on external validation (≈0.87-0.90) (Table 2; Figure 1). Traditional models (logistic regression, SVM) performed competitively but lagged by 0.04-0.10 absolute AUC depending on dataset. Deep learners and XGBoost were strong single baselines, aligning with prior evidence that representation-rich models excel on images and large tabular EHRs [1-6]. External AUCs were uniformly lower than internal AUCs (mean drop ≈0.02-0.03), reflecting well-known generalization challenges under dataset shift and hidden stratification [3, 11, 14].

Thresholded performance and clinical utility

At **90% specificity**, the Ensemble preserved the highest average sensitivity (≈0.69 external), followed by DNN and XGBoost (≈0.67 and ≈0.66). This operating region matches common early-warning preferences where false positives must be constrained to avoid alarm fatigue in clinical workflows [11-13]. On the MIMIC-AKI task representative of real-time EHR monitoring the Ensemble ROC dominated XGBoost (Figure 2) and exceeded it by ΔAUC≈0.02; DeLong tests confirmed statistical significance on MIMIC-AKI (p=0.006) and NIH CXR (p=0.012) but not on UCI Diabetes (p=0.08), suggesting task-dependent gains (Table 3) [11-13]. McNemar's test over pooled predictions showed fewer discordant errors for the Ensemble vs XGBoost (p=0.028), indicating a genuine improvement in classification decisions, not merely score re-ranking [11-13].

Calibration and reliability

Calibration curves (Figure 3) yielded slope≈0.93 and intercept≈0.02 on external validation with Brier score≈0.14, indicating clinically usable probability estimates after simple post-hoc calibration. This is consistent with recommendations that early-detection tools report both discrimination and calibration, per TRIPOD and CONSORT-AI/SPIRIT-AI standards [12, 13]. Good calibration is crucial where risk thresholds govern downstream actions (e.g., kidney-protective bundles for predicted AKI) [5, 6, 12, 13].

Robustness, imbalance, and subgroup behavior

Performance degrades modestly under class imbalance and between-site shift (Figure 1), but imbalance-aware training (sampling and cost-sensitive losses) preserved sensitivity in minority outcomes, in line with classic results on imbalanced learning [8]. Subgroup analyses (not shown) reflected hidden stratification patterns e.g., slightly lower AUCs in very elderly AKI patients and in atypical radiographic presentations—consistent with prior reports that aggregate metrics can mask clinically important failure modes [14, 3]. These findings reinforce the necessity of explicit subgroup evaluation and domain generalization checks before deployment [3, 11, 14].

Explainability and clinical face validity

SHAP analyses (Figure 4) highlighted Δcreatinine over 48h, urine output, baseline eGFR, age, and SOFA score as top contributors to AKI risk—variables with plausible clinical mechanisms—supporting face validity and aligning with explainability best practices (SHAP/LIME) [9, 10]. For imaging, analogous saliency-guided checks (not pictured) confirmed signal localization in pulmonary opacities rather than spurious confounders, a known risk in medical imaging models [2, 3, 14].

Alignment with reporting guidance

All analyses were conducted and reported in accordance with TRIPOD and CONSORT-AI/SPIRIT-AI recommendations (complete discrimination, calibration, external validation, and clear intended use) to enhance reproducibility and translational credibility [12, 13]. Together, these results substantiate the hypothesis that an interpretable, imbalance-aware Ensemble offers superior early-detection performance and reliability over single black-box baselines across modalities, while acknowledging remaining generalization constraints that mirror the broader literature [1-14].

Discussion

The findings of this study confirm that integrating diverse machine learning (ML) techniques—specifically ensemble and imbalance-aware models—can significantly enhance early disease detection performance across heterogeneous healthcare data sources. The superior results obtained by the ensemble approach, which outperformed all single models in AUC, calibration, and sensitivity, reinforce growing evidence that multi-model integration mitigates overfitting and leverages complementary decision boundaries [1, 3, 5]. Similar outcomes have been observed in prior research on skin cancer classification, pneumonia detection, and acute kidney injury (AKI) prediction, where ensemble and hybrid deep learning architectures improved diagnostic robustness and generalization [1-6].

A consistent performance gap between internal and external validation highlights the persistent dataset shift problem, where model performance degrades when applied to unseen populations [3, 11, 14]. This underscores the necessity for external, multicenter validation and recalibration before clinical deployment, a limitation repeatedly emphasized in systematic reviews of diagnostic ML models [7]. Despite robust internal metrics, clinical adoption requires reproducibility under real-world variability age distributions, disease prevalence, and sensor differences which often remain unaccounted for in academic studies [4, 11]. The present study's adherence to TRIPOD and CONSORT-AI/SPIRIT-AI reporting guidelines enhances methodological transparency, addressing previous concerns about incomplete reporting and unvalidated claims [12, 13].

The incorporation of explainable AI (XAI) tools such as SHAP and LIME further supports clinical interpretability, allowing clinicians to understand model reasoning at both global and local levels [9, 10]. The alignment of top predictive features Δcreatinine, urine output, and eGFR—with established AKI risk factors [5, 6] strengthens the clinical plausibility of the results and demonstrates that data-driven methods can recover mechanistic insights aligned with medical expertise. These findings are in line with previous calls for "interpretable-by-design" healthcare AI systems that combine predictive performance with transparency [9, 11].

Moreover, addressing class imbalance through synthetic oversampling (SMOTE) and cost-sensitive optimization preserved minority-class recall without excessively inflating false positives, consistent with foundational work on imbalanced learning [8]. The observed sensitivity-specificity balance meets practical thresholds for early-warning systems, which must prioritize timely alerts while minimizing alarm fatigue [11, 13]. Finally, the model's favorable calibration (Brier≈0.14) demonstrates clinically usable probability estimates, supporting risk stratification use cases rather than binary prediction alone [12, 13].

Overall, these findings validate the hypothesis that ensemble-based, interpretable ML frameworks trained on harmonized, multi-source data achieve superior accuracy and clinical reliability for early disease detection compared to individual black-box models. The study bridges the gap between theoretical AI research and deployable clinical systems by combining methodological rigor, interpretability, and generalizability key factors for future regulatory acceptance and real-world implementation in healthcare [1-14].

Conclusion

This study demonstrates that machine learning models, when thoughtfully designed and rigorously validated, hold transformative potential in the early detection of diseases across diverse healthcare domains. By integrating multiple algorithms through an ensemble approach and incorporating imbalance-aware training methods, the proposed framework achieved high discrimination power, robust calibration, and improved generalizability across heterogeneous datasets. The consistency of these outcomes across both clinical and imaging data confirms that predictive intelligence in healthcare must be built upon data diversity, interpretability, and continuous validation rather than relying solely on single, opaque models. The inclusion of explainable AI mechanisms, particularly SHAP-based feature attributions, ensured that clinical reasoning was transparent and aligned with established biomedical understanding, which strengthens the foundation for clinician trust and adoption. In practical terms, these insights provide actionable strategies for healthcare organizations and researchers. Hospitals and health systems should prioritize the development of ML pipelines that combine structured EHR data, medical imaging, and laboratory results to create unified prediction systems that can detect disease onset well before clinical manifestation. Moreover, clinical researchers should embed imbalance-aware sampling and cost-sensitive optimization into model design to prevent systematic underperformance in rare but critical subpopulations. To promote reliability, routine recalibration and external validation across multi-institutional datasets should become a standard practice before clinical deployment. In operational settings, explainable models should be integrated into decision support tools in ways that assist rather than replace clinician judgment, ensuring ethical and accountable AI use. Policymakers and hospital administrators should also establish dedicated AI governance frameworks that define protocols for data quality, bias mitigation, and real-time model monitoring. For continued progress, interdisciplinary collaboration between clinicians, data scientists, and regulatory authorities must be strengthened to align technical innovation with patient safety and public health objectives. Future research should focus on expanding data interoperability standards, building domain-adaptable models that maintain stability across diverse demographics, and integrating continuous learning mechanisms that update model parameters as new data become available. Collectively, these measures will accelerate the translation of machine learning from experimental environments to everyday clinical practice, empowering healthcare systems to detect and manage diseases at their earliest, most treatable stages while upholding transparency, fairness, and human-centered care.

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