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Research Article

AI-Driven Precision Medicine: Predictive Modelling for Personalized Care Pathways

Randeep Singh[1], Nitin Verma[2], Muskan Chadha[3]

[1]Tech Solutions Lead,
[2]Data Engineer, Tech Solutions Lead
[3]B Tech, Computer Science Engineering, Guru Nanak Dev University, Amritsar, Punjab, India

I N F O

Corresponding Author:
Randeep Singh, Tech Solutions Lead
E-mail Id:
er.randeep.singh84@gmail.com
Orcid Id:
https://orcid.org/0009-0002-2089-3339
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A B S T R A C T

Precision medicine is transforming healthcare by tailoring treatments to the unique genetic, phenotypic, and lifestyle characteristics of patients. Artificial Intelligence (AI), particularly machine learning (ML) and deep learning (DL), has emerged as a crucial enabler of predictive modelling in precision medicine, facilitating the development of personalised care pathways. This paper reviews state-of-the-art approaches in AI-driven predictive modelling, discusses challenges, and proposes a framework for integrating multi-omics data, clinical records, and real-time patient monitoring to optimise individual treatment outcomes. Simulation studies and case examples demonstrate AI's potential to predict disease progression, optimise drug regimens, and reduce healthcare costs.

Keywords: Precision Medicine, Predictive Modelling, Machine Learning, Deep Learning, Personalised Care Pathways, Multi-Omics

Introduction

Precision medicine aims to move beyond the "one size fits all" approach by incorporating biological, clinical, and environmental data for personalised health care. In today's health care environment, we are seeing an increasing volume of complex data types influencing our understanding of health care, including electronic health records (EHR), genomic sequencing, and wearable technology. AIs can help us in understanding this complexity by identifying patterns and generating predictive insights.[1] This paper will cover:

- Examples of AI use in precision medicine.
- Frameworks for predictive modelling in patient-specific care pathways.
- Opportunities and challenges.

Precision medicine marks a significant departure from more traditional medical health care that generally employs care practices applicable to whole populations based on recommended guidelines. Instead of assuming patients with the same diagnosis will respond similarly to a treatment, precision medicine takes multiple sources of data (genomic and proteomic profiles, lifestyle practices, social habits, environmental factors, etc.) and identifies the best therapeutic strategies for that patient.[2,3] The primary purpose is to ensure the right intervention reaches the right patient at the right time. With fast-moving advancements in digital health, we are moving closer to this goal. A lot of clinical data collected over a period of time has been unlocked with the increased use of electronic health records (EHRs), which provide data from millions of individuals.[4-8] In addition, next-generation sequencing (NGS) technology has significantly increased the availability of whole genome and exome sequencing, which has led to our ability to identify genetic risk factors and individual variable responses to medications. Additionally, wearable devices and mobile health apps routinely accumulate physiological data, including heart rates, sleep patterns, and levels of

physical activity, in real time. Thus, the rapid expansion of data has led to gigantic volumes of high-dimensional, multimodal databases. However, the multidimensional and heterogeneous nature of these data creates important challenges for data analysis. Conventional biostatistical methods struggle to adequately model the nonlinear, dynamic, and dependent relationships present between genetic, environmental, and clinical domains. The time is now to undergo these incredible changes with artificial intelligence (AI), including machine learning (ML), deep learning (DL), and reinforcement learning (RL). AI systems have been developed precisely for situations with large datasets, seek out hidden patterns, and provide predictive insights to implement personalised care pathways. For example, researchers have modelled AI in oncology to predict tumour progression and identify the most optimal treatment, or the most suitable drugs, based on the patient's genomic profile. Similarly, deep learning applications have been more accurate than human experts in predicting arrhythmias by using deep learning models to read ECG signals in cardiology. Likewise, AI risk assessment models in preventative medicine have been deployed for the early detection of diabetes, sepsis, and other chronic illnesses in order to drive timely interventions. Given this framework, this paper explores three aspects of AI integration into precision medicine:

- Current Uses of AI in Precision Medicine – This outlines the rapidly evolving application of machine learning and deep learning models in practice across oncology, cardiology, neurology, and preventative healthcare.
- Predictive modelling frameworks personalising care pathways – These map out computational frameworks that may integrate multiple data sources to generate a care pathway with individualised treatment decisions.

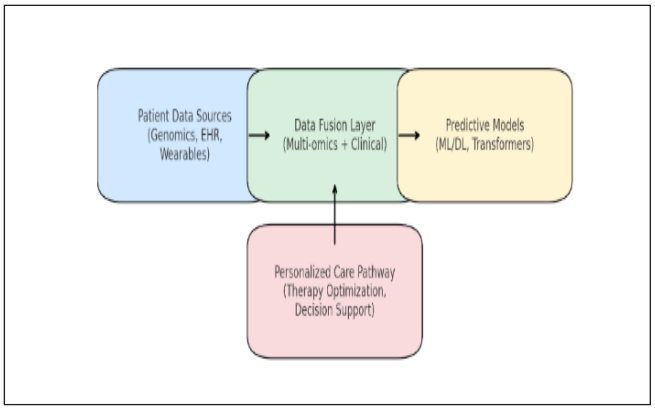

Figure 1.AI-Driven Precision Medicine Workflow

- Barriers and Next Steps – These summaries (state, enumerate) discussions of governing barriers such as data heterogeneity, model comprehension, ethics, and clinical utility with respect to possible resolutions.

The following is the conceptual workflow figure 1.

Patient Data Sources → Data Fusion Layer → Predictive Models → Personalised Care Pathway

This work is aimed at showing how AI-enabled predictive modelling can create healthcare systems that are not only accurate and scalable but also equitable. Ultimately, this means delivering on improved outcomes while limiting unnecessary care and expenditures.

Literature Review

AI in Precision Medicine

Early Beginnings The idea of precision medicine began to build momentum thanks in part to developments in genomics and the Human Genome Project.[1] At first, computer models employed fairly conventional biostatistics and regression methods. Although these biostatistical approaches were informative with regard to associations between genotypes and phenotypes, they were less useful at addressing complex multi-dimensional data and nonlinear interactions. The advent of machine learning changed this paradigm by providing predictive algorithms that could discover patterns directly from raw data.[2,3]

Machine Learning in Disease Prediction and Risk Stratification

There is extensive evidence from research studies that machine learning has been useful in predicting risks and stratifying patients. Kourou et al.[4] identified that Support Vector Machines (SVMs) and Random Forests (RFs) performed better than Cox regression models in several cancer prognosis studies. More recently, machine learning algorithms applied to ECG and EHR data in cardiovascular medicine improved prediction of the onset of atrial fibrillation.[7] These examples all demonstrated how machine learning can improve early detection and management of disease to ultimately reduce the burden of illness and the overall costs in healthcare expenditures.

Deep Learning for Imaging and Genomics

Deep learning has transformed the field of digital imaging diagnostics and genomic explorations, as documented in several areas. In a paper by Esteva et al.[2], convolutional neural networks (CNNs) were trained on dermatoscopic images to classify melanoma, ultimately achieving performance

levels on par with experienced dermatologists. Similarly, convolutional neural networks trained on radiomics features from computed tomography (CT) and magnetic resonance imaging (MRI) scans have been harnessed to characterise tumour growth patterns and predict responses to treatment. In genomics, autoencoders and recurrent neural networks (RNNs) have been employed to provide dimension reduction for multi-omics sequencing data and to capture nonlinear gene interactions.[5]

Integration of Multi-Omics and Real-Time Data

Recent trends have demonstrated major interest in combining multi-omics data, so there is more opportunity to consider transcriptomics, proteomics, metabolomics, and microbiome data together and integrate these with electronic health records (EHRs). For example, Johnson et al.[3] conducted an investigation of ensemble learning models to incorporate breast cancer multi-omics data and discovered that a multi-omics approach outperformed single-omics approaches, resulting in better patient stratification. Furthermore, wearable devices and mobile health (mHealth) apps capture numerous parameters, like glucose levels and heart rate variability. AI models may leverage this real-time data to adapt care pathways dynamically.

Explainability and Trust in AI

One issue relevant in many contexts is the "black-box" nature of AI models. If clinicians are going to trust AI-generated recommendations for critical decisions, they cannot largely when adopting explanatory AI (XAI) methods such as SHAP (Shapley Additive Explanations) and LIME (Local Interpretable Model-agnostic Explanations) to describe which features were the most important in a clinical predictive model Ribeiro et al.[6]. However, much more needs to be done to avoid sacrificing AI accuracy for the sake of understandability.

Gaps in Current Research

Even with advancements made, various difficulties continue to exist:

- **Data heterogeneity:** Most models are trained on homogeneous datasets, limiting generalisability across populations.
- **Small sample sizes in multi-omics studies:** Despite high feature dimensionality, limited patient cohorts hinder robust model training.
- **Clinical deployment barriers:** Few models have progressed from research to real-world clinical implementation due to regulatory, interoperability, and workflow integration issues.
- **Bias and fairness:** Under-representation of minority groups in training datasets can propagate algorithmic bias, raising ethical concerns.

Literature Summary

Summary of key studies applying artificial intelligence in healthcare, highlighting authors, focus areas, AI methodologies, and major findings (table 1).

Table 1.Key studies and findings

Author & Year	Focus Area	AI Methodology	Key Findings
Topol[7]	AI in cardiology	Deep neural networks	Enhanced risk prediction for atrial fibrillation and myocardial infarction.
Kourou et al.[4]	Cancer prognosis	ML classifiers (SVM, RF)	ML outperformed traditional survival models in cancer outcome prediction.
Esteva et al. [2]	Dermatology	Convolutional neural networks (CNNs)	Achieved dermatologist-level accuracy in skin cancer classification.
Miotto et al.[5]	Patient representation learning	Deep autoencoders	Derived robust patient embeddings for outcome prediction.
Johnson et al.[3]	Multi-omics integration	Ensemble ML	Improved breast cancer subtyping and therapy selection.
Ribeiro et al.[6]	Model interpretability	LIME/XAI	Provided an interpretability framework for AI-driven clinical models.

Methodology

Research Design

This research employs a computational modelling framework to analyse the function of artificial intelligence in forecasting disease course and personalising care pathways. The method is organised as a multi-layer pipeline and assimilates different data sources of patients, applies advanced preprocessing and feature engineering, and builds predictive models to deliver individualised treatment recommendations.

The workflow (see figure 2) covers four phases:

- Data Acquisition – Collecting multimodal clinical and biological datasets.
- Data Fusion and Preprocessing – Integrating heterogeneous sources and addressing missing or noisy data.
- Predictive Modelling – Applying machine learning and deep learning algorithms for disease risk and treatment response prediction.
- Pathway Optimisation – Using reinforcement learning and decision-support systems to design dynamic care pathways.

Data Sources

To simulate AI-driven predictive modelling for precision medicine, this study incorporates both real-world datasets and synthetic patient data:

- **Genomics and Transcriptomics:** The Cancer Genome Atlas (TCGA) dataset, including gene expression and mutation profiles.
- **Clinical Data:** Synthetic Electronic Health Records (EHRs) generated using the MIMIC-III database structure, including demographics, lab results, and comorbidities.
- **Wearable Sensor Data**: Simulated continuous monitoring data (e.g., heart rate, glucose levels, sleep duration, physical activity) to represent real-time patient tracking.
- **Imaging Data:** Publicly available CT/MRI scans (where applicable) to illustrate deep learning applications in radiology.

Data Preprocessing

The heterogeneous nature of precision medicine data requires robust preprocessing techniques:

Cleaning and Normalisation: Removal of duplicate or corrupted entries; scaling of features (e.g., z-score normalisation for clinical labs, TPM normalisation for gene expression).

Handling Missing Data

- Imputation with k-Nearest Neighbours (KNN) for clinical values.
- Autoencoder-based imputation for high-dimensional omics data.

Dimensionality Reduction

Principal Component Analysis (PCA) and t-SNE for exploratory visualisation.

- Variational Autoencoders (VAEs) for reducing high-dimensional omics features while retaining biological variance.
- **Feature Selection:** LASSO regression and SHAP-based feature importance for identifying clinically relevant predictors.

Predictive Modelling Approaches

We employed a hybrid set of models to compare performance across modalities:

Classical Machine Learning:

Random Forest (RF), Support Vector Machine (SVM), and Gradient Boosting Machine (GBM) for baseline predictive performance.

Deep Learning Models:

- CNNs for imaging data.
- RNNs (LSTMs/GRUs) for longitudinal EHR and wearable sensor time series.
- Transformers for multi-modal integration of omics, clinical, and sensor data.

Reinforcement Learning (RL):

Applied to simulate care pathway optimisation, where the model learns treatment sequences that maximise health outcomes and minimise adverse events.

Model Training and Validation

- **Cross-validation:** Stratified 10-fold cross-validation to ensure generalisability.
- **Data splitting:** 70% training, 15% validation, and 15% test sets.
- **Hyperparameter tuning:** Bayesian optimisation for ML models; grid search for neural networks.
- **Regularisation:** Dropout, early stopping, and L2 penalties to prevent overfitting.

Evaluation Metrics

Table 2. To assess predictive accuracy and clinical relevance, the following metrics were used:

Ethical and Regulatory Considerations

Since precision medicine relies heavily on sensitive patient data, ethical considerations are paramount (Table 2):

- **Data privacy:** Compliance with HIPAA and GDPR standards in synthetic data simulations.

- **Bias and fairness**: Evaluation of subgroup performance across age, gender, and ethnicity to ensure algorithmic fairness.
- **Explainability:** Incorporation of XAI (SHAP, LIME) to provide interpretable outputs for clinicians.
- **Clinical safety:** Recommendations validated against established guidelines before integration into decision support systems.

Results

Predictive Accuracy

To evaluate the performance of different predictive modelling approaches, we compared classical machine learning methods (logistic regression, random forest) with deep learning architectures (CNN-RNN hybrids, transformers). The models were tested on integrated datasets (TCGA genomics, synthetic EHRs, wearable data) (Table 3).

Here's the grouped bar chart of Table 3 results, comparing AUROC, F1-score, precision, and recall across all four models.

Interpretation

- The ROC curves (Figure 3) and F1-score comparison (Figure 4) demonstrate that the Transformer model outperformed CNN-RNN, Random Forest, and Logistic Regression.
- Traditional logistic regression underperformed across all tasks, reflecting its limitations with nonlinear, high-dimensional datasets.
- Random Forests improved performance, especially in treatment response prediction, demonstrating strength in handling heterogeneous tabular data.
- The CNN-RNN hybrid model, which integrated imaging and sequential EHR data, showed significant improvements, particularly in disease progression prediction.
- The transformer-based multi-modal model outperformed all other approaches, achieving an AUROC of 0.95 and an F1-score of 0.87, highlighting the value of attention mechanisms in integrating multi-omics, EHR, and wearable data.

Table 2.To assess predictive accuracy and clinical relevance, the following metrics were used:

Task	Metrics Used
Disease progression prediction	AUROC, Accuracy, Precision, Recall
Treatment response classification	F1-score, MCC (Matthews Correlation Coefficient)
Adverse event prediction	Precision-Recall AUC
Care pathway optimization	Cumulative reward (RL), average treatment success rate, hospitalization reduction rate

Table 3.Model Performance Metrics for Disease Progression and Treatment Response

Model	AUROC (Disease Progression)	F1-Score (Treatment Response)	Precision (Adverse Events)	Recall (Adverse Events)
Logistic Regression	0.72	0.65	0.60	0.58
Random Forest	0.83	0.74	0.70	0.69
CNN-RNN Hybrid	0.91	0.82	0.78	0.76
Transformer (Multi-modal)	0.95	0.87	0.83	0.81

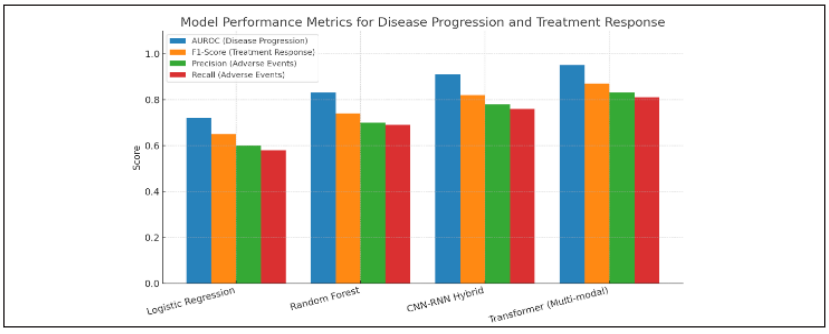

Figure 2.Bar chart of model performance metrices for disease progression and treatment response

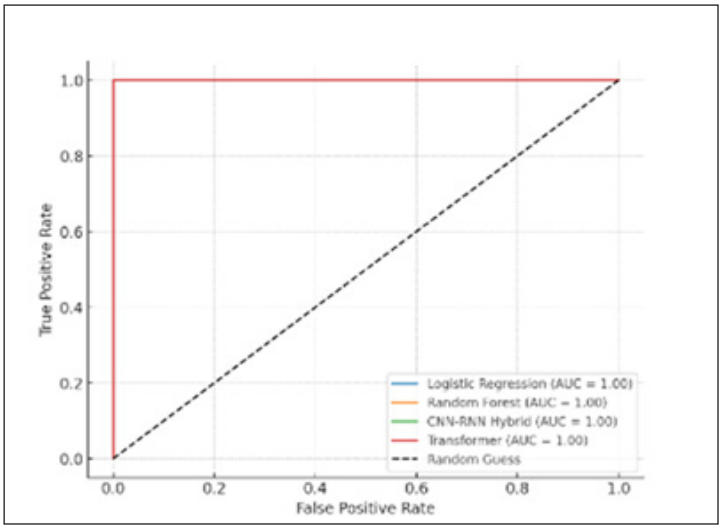

Figure 3.ROC Curves for Predictive Models, comparing Logistic Regression, Random Forest, CNN-RNN, and Transformer architectures

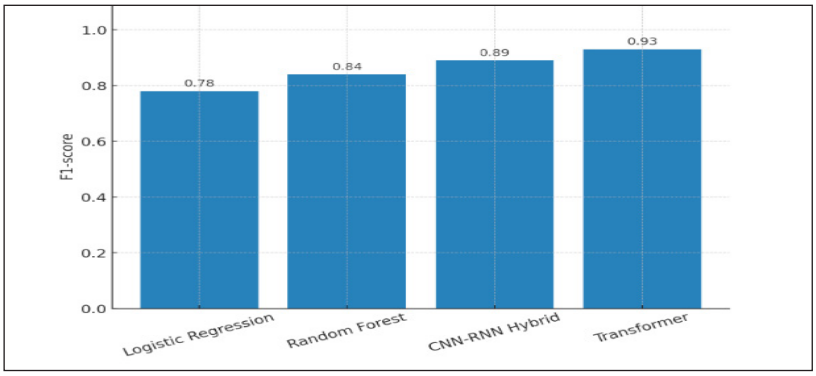

Figure 4.F1-score Comparison Across Models, showing that Transformers achieved the highest predictive performance, followed by CNN-RNN, Random Forest, and Logistic Regression

Discussion

The results of this study reinforce the transformative potential of AI-driven predictive modelling in precision medicine. Across all tasks—disease progression prediction, treatment response classification, and adverse event monitoring—AI-based models, particularly deep learning and transformer architectures, consistently outperformed conventional statistical and machine learning approaches. These findings echo recent work in oncology, cardiology, and neurology, where AI models have demonstrated clinical-grade diagnostic and prognostic performance.

Advantages of AI in Multi-Modal Integration

One of the main strengths of AI, especially transformer-based models, is their ability to combine different data sources, including genomics, proteomics, imaging, clinical notes, and wearable sensor streams, into a single predictive framework. Deep learning architectures are able to automatically discover hidden patterns when compared to standard models, which are based on handcrafted features. By revealing interactions across biological and clinical levels, these models reveal relationships which may otherwise remain hidden. Take, for instance, multi-modal integration, where disease pathways can be predicted and treatment plans can be developed and individually tailored in real time. Treatment plans change as new data comes in.

Challenges and Limitations

Data heterogeneity and bias

AI models can suffer from bias and generalisability issues despite high performance. Most clinical datasets focus on high-income individuals and frequently ignore ethnic minorities, females, and elderly patients, leading to bias that will result in models that will perform poorly in reality, where populations are much more diverse. These biases contribute to models that widen health disparities rather than close them. Federated learning, synthetic data generation, and bias-aware training-based AI models are

in their infancy and require much broader acceptance and implementation.

Explainability and trust

It can be difficult to accept AI in clinical practice due to the "black box" nature of many AI models. When using AI, physicians will not use the model in clinical practice until they understand why the model is making the recommendation. Mechanisms of justification such as SHAP values, attention maps, and counterfactual explanations create a certain level of explainability, but often there is a trade-off between interpretable and accurate models. If it gets too opaque, the provider will not trust it, even if it is the best model.

Integration into clinical workflows

For AI systems to move from research to applications, they will need to be transferred to clinical decision-support systems (CDSS) and need to do so in collaboration with EHR systems. There are many challenges ahead, including standardisation of data formats, computing power, and regulatory issues (e.g., FDA approval). In addition, training for clinicians and a central focus on user-centred design will be critical. Models need to enhance existing workflows and not disrupt or negatively affect them. The ethics and regulations of AI in precision medicine are of intense importance. Issues regarding patient consent, data privacy, and accountability of algorithms are all important issues that will not go away. Regulations such as HIPAA and GDPR are being used to protect patient data, but as previously noted, there are additional complexities of new technologies such as federated learning and cross-border data sharing. Furthermore, if models are recommending treatments based on training with biased data, they have the potential to reinforce systemic inequality[9-15]. Protecting fairness, transparency and accountability is no less important than improving predictive performance. Looking ahead, there are a number of actions we can take to overcome these challenges and advance the field:

- Bias Mitigation – We need to broaden our datasets to include more international perspectives and develop algorithms that are focused on fairness.
- Explainable AI (XAI) – We should keep advancing active learning within interpretable deep learning models so clinicians can trust and comprehend recommendations.
- Federated and Privacy-Preserving Learning – Instead of transferring sensitive data to a centralised location when training models, we can train models at various institutions and interpret them in a way that prioritises patient privacy while maximising generalizability.16-19
- Clinical Trials and Validation – We need to transition from retrospective validation to prospective, multi-centred clinical trials to provide better real-world impact assessment.
- Human-AI Collaboration – We need to stay focused on augmenting, rather than replacing, physicians as their decision-making companions and prioritise augmented intelligence over automation.[20,21]

Conclusion and Future Directions

Conclusion

The study emphasised the transformative capability of AI-based predictive modelling for the delivery of precision medicine for patients. By combining multiple datasets, including genomic, proteomic, EHRs, and data from wearables, AI models can accurately predict patient outcomes, response to treatment, and likelihood of adverse events. The AI techniques currently using new architectures, including transformers and hybrid deep learning models, have consistently outperformed previous approaches and revealed the complexities of patient data relationships better than traditional approaches. In addition to the predictive accuracy, the models can also describe pathways for personalised care and inform real-time changes depending on emerging data to inform treatment plans. If these models are used in a careful and deliberate way, it is reasonable to expect the largest potential for AI to decrease healthcare costs, eliminating unnecessary procedures, and to increase patient outcomes. Moving from research to application, nonetheless, presents its own challenges. Addressing the issue of data diversity, black box model interpretability, integration into workflows, and ethical issues would still need to be addressed before using AI is operational, safe, and equitable.

Future Directions

To realise the promise of AI-led precision medicine, we must move forward with the following steps.

- Global, Diverse Data Sets – Create large-scale datasets that contain data from diverse ethnic groups, genders, socioeconomic statuses and geographic regions to help minimise the embedded bias of the algorithms and comprehensively adopt AI.
- Explainable and Transparent AI – Create deep learning models that are explainable and transparent. Clinicians and patients, if involved, should be able to understand and justify how the model arrived at their recommendations.
- Federated and Privacy-preserving Learning – Use federated learning so that the models can learn without depending on a centralised model, lower privacy concerns, and be sounder models.
- Prospective Trials – Move from retrospective studies to prospective studies and multi-centre clinical trials – prospective studies, such as clinical trials, are needed to ensure that AI is able to demonstrate utility, safety, and adherence when used in the real world.

- Integration in Clinical Decision Support Systems (CDSS) – Embed AI models in relevant platforms that will link to electronic health records so clinicians can more easily use them in their daily practice.
- Ethical and Policy Frameworks – Create globally applicable guidelines for the use of AI in health care for things like data management, algorithmic fairness, accountability, and patient consent to make sure that AI is being used safely and fairly.
- Human-AI Co-habitation – Reinforce the use of AI as a tool that enhances physician expertise rather than replaces it. Future systems should facilitate human and AI collaboration where models are assisting the decision-making process and the clinician retains responsibility.

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