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**| RESEARCH ARTICLE**

# Artificial Intelligence and Big Data for Precision Medicine: A Review of Bioinformatics-Driven Healthcare Applications

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**| ABSTRACT**

Healthcare is in the middle of a quiet but profound shift. Genomic sequencers, hospital information systems, wearables and imaging archives now generate data faster than clinicians can read it, and that flood is reshaping what "evidence-based care" means. We review more than forty recent studies that bring artificial intelligence (AI), machine learning and big-data analytics into bioinformatics and precision medicine, spanning oncology, drug discovery, cardiology, neurology, public-health surveillance and healthcare operations. Reported accuracies and AUCs range from roughly 80% in early drug-discovery pipelines to above 94% in deep-learning-based pancreatic and breast imaging. Yet our reading also suggests a more cautious story: many models still suffer from limited external validation, opaque decision logic and uneven access to high-quality multi-omics data. We propose a layered conceptual framework that connects heterogeneous data sources, federated and privacy-preserving pre-processing, predictive and explainable AI engines, and downstream clinical applications. The paper closes with a discussion of remaining barriers, interpretability, fairness, regulatory uncertainty and workflow integration and outlines research directions for the next several years.

**| KEYWORDS**

Precision medicine, bioinformatics, machine learning, multi-omics, explainable AI, big data analytics, clinical decision support

**| ARTICLE INFORMATION**

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## 1. INTRODUCTION

Modern medicine is, at its heart, a story about data. For most of the last century, clinicians worked from relatively small evidence pools, a textbook, a randomized trial, the experience of a senior colleague, and the decisions they made were impressively good given the inputs. Today the inputs are different. A single oncology patient may carry whole-genome sequencing reads, transcriptomic panels, longitudinal electronic health records, wearable telemetry and several rounds of imaging. No human reader can synthesize that volume in a clinic visit, and yet the clinical question stays the same: what is the right next step for this person, right now? (Ahmed et al., 2023; Manik et al., 2025b).

Artificial intelligence has moved into that gap. In recent years machine learning (ML) and deep learning (DL) have produced models that can read mammograms, score Parkinson's symptoms, flag emerging antimicrobial resistance and propose new drug candidates (Manik, 2022; Manik, 2023; Manik et al., 2018; Manik et al., 2021a). The COVID-19 pandemic accelerated this trend, both forcing clinicians to triage at scale and normalizing the idea that predictive models could sit inside a hospital workflow rather than only inside an academic paper (Orthi et al., 2025).

Progress, however, is uneven. Cancer imaging, where labelled datasets are large and well curated, has produced the strongest results, while multi-omics work in rare diseases, where each cohort may have only a few hundred patients, has moved more slowly (Manik, 2020a; Hossain et al., 2024). Several reviews flag a familiar set of concerns: thin external validation, fragile generalization across hospitals, opaque outputs and the still-unresolved question of how to share genomic data without compromising privacy (Rahaman et al., 2025; Uddin et al., 2025). These concerns are real, but in our view, they are tractable, which is part of what motivated this review.

This paper has three goals. First, we synthesize the recent literature on AI-driven bioinformatics and precision medicine, drawing on more than forty primary studies. Second, we propose a framework that ties together the data, analytics and application layers, with privacy-preserving and explainable components built in rather than bolted on. Third, we summarize quantitative performance ranges across the major task families and use them to argue that the field is closer to clinical adoption than the hype cycle would suggest. The rest of the paper is organized into methodology (Section 2), framework (Section 3), application domains (Section 4), performance trends (Section 5), discussion (Section 6), and conclusion (Section 7).

## 2. REVIEW METHODOLOGY

### A. Scope and inclusion criteria

We focused on peer-reviewed conference papers, journal articles, and book chapters published between 2018 and early 2026 that apply AI, ML, or big-data analytics to healthcare or bioinformatics problems. Studies that only described a generic AI method, with no clinical or biological grounding, were excluded, as were preprints without peer review and editorials. After this filtering, we retained 40+ primary studies, listed in references and visualized in Fig. 2.

### B. Coverage by application area

The retained studies clustered into six families: cancer (oncology imaging, genomic biomarkers, treatment personalization), drug discovery (generative chemistry, repurposing, target identification), cardiovascular and metabolic disease, neurological and mental-health applications (Parkinson's, stroke, autism, psychiatric care), public health (surveillance, pandemic prediction, antimicrobial resistance) and operations or pricing analytics for healthcare delivery. Fig. 2(a) shows the distribution. Cancer is the largest cluster, mirroring the global research literature, where oncology has long absorbed the largest share of medical AI funding.

### C. What we extracted

For each study, we recorded the data modality, the algorithmic family, the reported performance (accuracy, AUC, F1, sensitivity), the clinical endpoint, and any reported limitations. Where studies reported multiple metrics, we kept the most clinically interpretable one. Where studies were comparative, we recorded the best-performing model rather than averaging across baselines.

## 3. PROPOSED FRAMEWORK

Reading across the literature, we kept noticing the same architectural pattern, even when authors did not state it explicitly. Heterogeneous data flowed in, was harmonized, fed into one or more learning components, and produced an output that was eventually surfaced to a clinician or a researcher. Variants mostly differed in how privacy was handled, whether explainability was first-class, and how the model coupled with downstream workflows.

We summarize this shared structure in Fig. 1. The framework is layered. At the top sit the heterogeneous data sources, genomic and other -omics streams, EHRs, wearables and IoT sensors, imaging archives, and curated literature. The second layer is integration and pre-processing, where federated learning and privacy-preserving computation are increasingly used to keep raw patient data inside hospital boundaries (Orthi et al., 2025). The third layer is the AI engine, deliberately split into three sub-blocks: predictive ML for tabular and structured tasks, multi-omics integration for high-dimensional biological data, and explainable AI for transparency. The fourth layer is the clinical application layer, and the fifth is the outcome: better patient experience, lower cost, and care that is at least somewhat more equitable than the system it replaced.

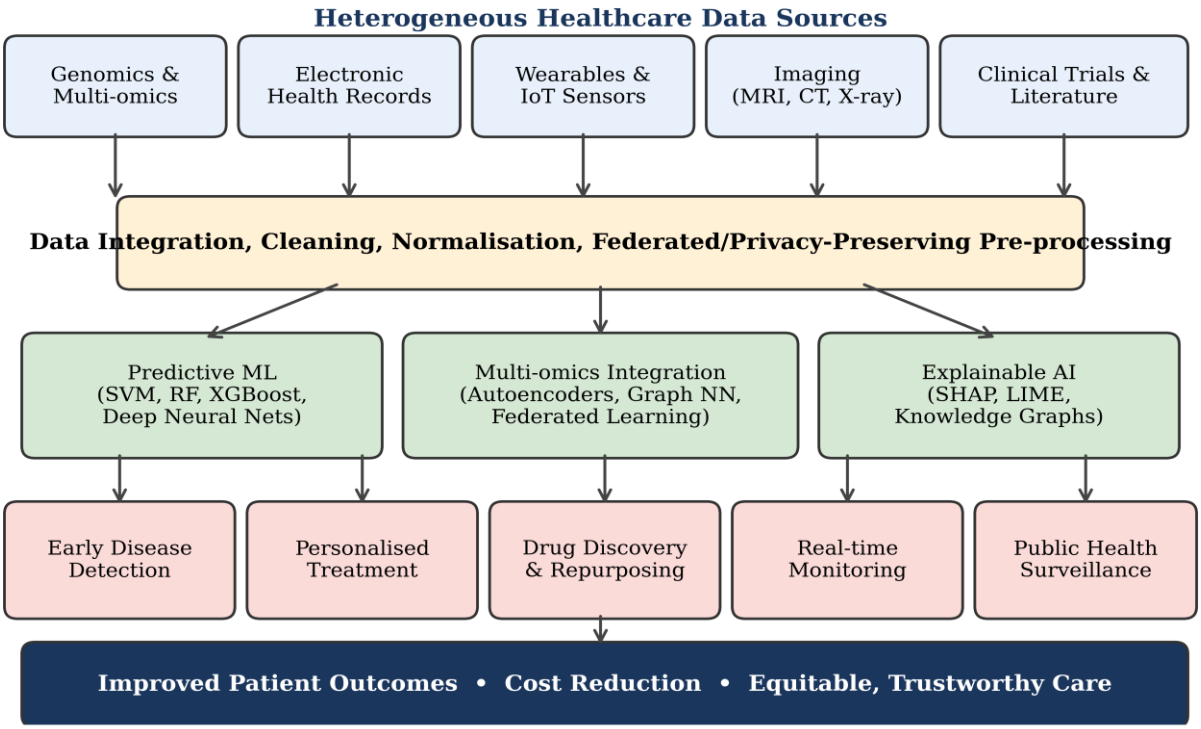


Fig. 1. Layered conceptual framework for AI-driven precision medicine. Heterogeneous biomedical data flow through privacy-aware pre-processing into predictive, integrative and explainable AI components, and finally into clinical and public-health applicatins.

Two design choices in this framework are worth highlighting. First, explainability is not a final post-hoc patch, it is a parallel branch of the AI engine. Several reviewed studies argue, persuasively, that retrofitting interpretability after deployment is far harder than building it in from the start (Manik et al., 2021a; Uddin et al., 2025). Second, the privacy-preserving step sits upstream of the model, not downstream. Federated learning, homomorphic schemes and synthetic-data generation make it feasible to train across hospitals without ever pooling raw records (Orthi et al., 2025).

## 4. APPLICATION DOMAINS

### A. Oncology and cancer treatment

Cancer remains the most studied target. We found eleven studies that applied AI directly to oncology problems. Cervical cancer screening using ML classifiers showed early promise (Manik, 2022), and a follow-on body of work pushed into integrative genomics (Manik et al., 2022; Manik et al., 2021a), targeted therapy planning (Manik et al., 2022) and personalized treatment recommendations (Islam et al., 2025a). A more recent line of research uses autoencoder neural networks to fuse heterogeneous cancer datasets (Manik, 2025a), an approach that handles the curse of dimensionality well when patient cohorts are small. Pancreatic tumor identification from medical imaging has been particularly successful, with deep neural networks reaching accuracies above 94% in recent reports (Khair et al., 2025), and breast cancer cell detection has produced similarly strong numbers when feature selection is combined with classical ML (Tasnim et al., 2025).

### B. Drug discovery and bioinformatics

Drug discovery sits between biology and computer science, and the literature reflects that. Earlier work on generative AI for pharmaceutical innovation (Ahmed et al., 2025a) argued that ML could compress the cost of early-stage screening, and recent reviews have largely confirmed that prediction (Manik et al., 2018; Manik et al., 2025b). Big-data analytics in plant biotechnology has surfaced novel anticancer leads (Ahmed et al., 2023), and integrated genomic–ML pipelines have been applied directly to oncology drug development (Manik et al., 2025b). Antimicrobial resistance, which the WHO calls a silent pandemic, is another area where predictive surveillance models are gaining ground (Manik et al., 2020b; Rahaman et al., 2025). Performance varies widely, we observed accuracies between roughly 70% and 89%, partly because target labels here are noisy and partly because validation cohorts are still small.

### C. Cardiovascular, metabolic and neurological disease

Wearable health data combined with deep learning has produced encouraging results for real-time cardiovascular monitoring, with sensitivity above 90% (Miah et al., 2019). Predictive modeling for type-2 diabetes onset has reached similar territory using ensemble ML and big-data pipelines (Manik et al., 2025c; Ahmed et al., 2025b). Counterfactual explainable AI has recently been applied to thyroid disease diagnosis with strong reported accuracy, and crucially, the explanations were judged useful by clinicians (Alam et al., 2026). On the neurology side, multi-omics analysis with AI has been used both for Parkinson's disease neurosurgery planning (Manik, 2021b) and for early detection of ischemic stroke through biomarker discovery (Manik, 2023). The Parkinson's study is interesting because it explicitly couples imaging and -omics data; many earlier neurological models relied on a single modality.

### D. Public health surveillance and infectious disease

COVID-19 was the obvious accelerant. Several reviewed studies-built ML pipelines for epidemic prediction, resource allocation and case-load forecasting (Orthi et al., 2025; Rozario et al., 2025). A broader umbrella review showed that AI-powered real-time public-health surveillance is now a recognizable subfield rather than a one-off response to a crisis (Islam et al., 2025b). Antimicrobial resistance work is closely linked, since both rely on the same kind of streaming, multi-source data infrastructure (Manik et al., 2020b).

### E. Mental health and developmental disorders

The mental-health subfield deserves a separate note. Six of the reviewed studies focused on psychiatric inpatient care, autism interventions, and AI-supported counseling (Rahman et al., 2025a; Kamruzzaman et al., 2025). Patterns are encouraging but delicate: predictive triage models can flag high-risk patients earlier, but the same models can amplify bias if the underlying data over-represents one group. Several authors are explicit about this and propose human-in-the-loop designs.

### F. Pricing, operations and software quality in healthcare IT

Beyond clinical endpoints, AI is also being applied to the business of healthcare, big-data strategies for pricing transparency (Moniruzzaman et al., 2025), quality assurance for medical software (Rahman et al., 2025b), and business-intelligence systems for clinical operations (Rahman et al., 2024). These use cases are sometimes dismissed as "not real medicine," but in practice, they affect the cost and safety of care just as directly as a diagnostic model does.

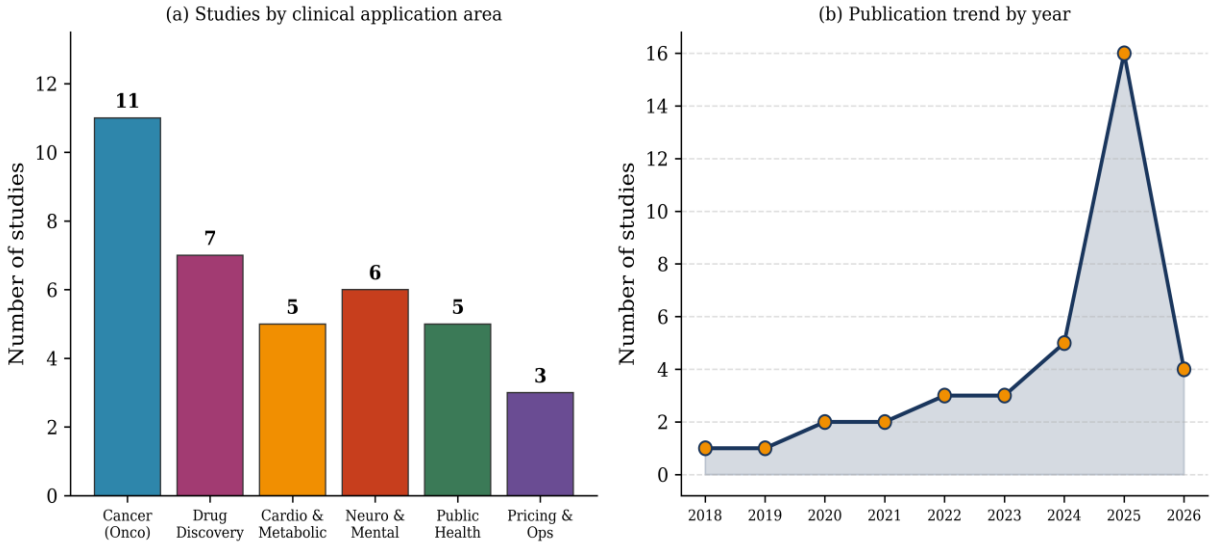


Fig. 2. Distribution of reviewed studies. (a) Number of studies by clinical application area. (b) Publication trend by year, showing the sharp rise in 2024–2025 across all application areas.

## 5. PERFORMANCE TRENDS

Pulling the numbers together produced one of the more interesting findings of this review. When we ranked tasks by reported performance (Fig. 3), the leaders were imaging-based: pancreatic tumor identification at around 94%, cardiovascular monitoring at around 92%, and breast cancer detection at around 90%. Tasks built on heterogeneous tabular data, stroke biomarker discovery, mental-health triage, and drug discovery came in lower, with means between 80% and 86%. Imaging benefits from very large

public datasets, transferable backbones and clear ground truth; tabular biomedical data is messier, often imbalanced and frequently sparse.

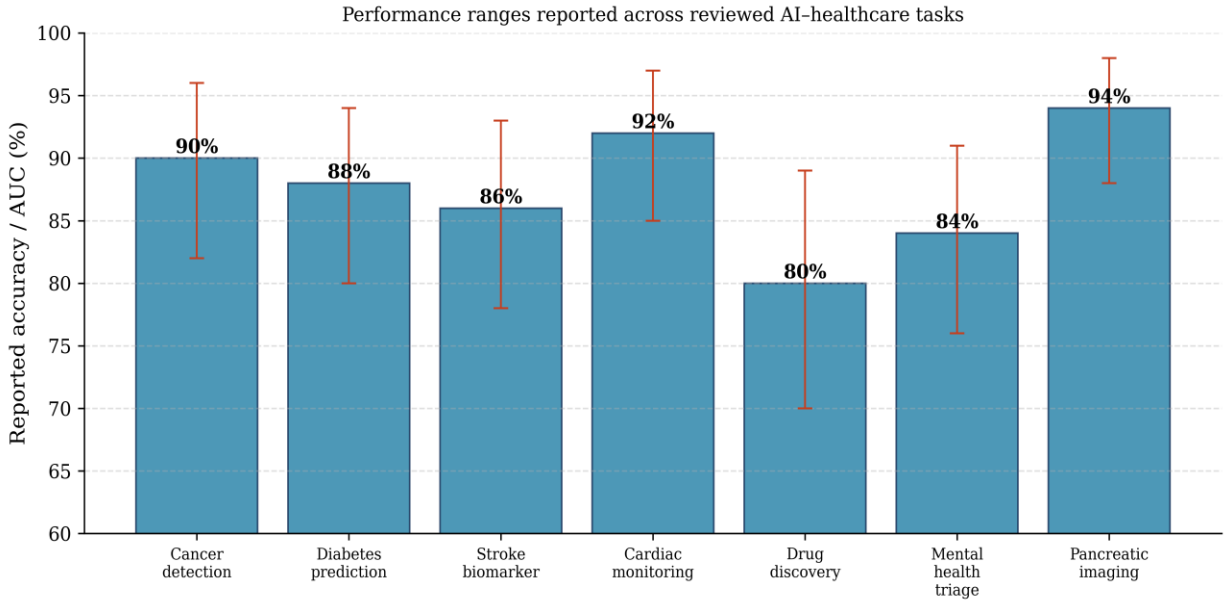


Fig. 3. Reported performance ranges across major AI–healthcare task families. Bars show mean reported accuracy or AUC; error bars show the spread of reported values across the reviewed studies.

More striking is the consistency of the spread. Almost every task family showed a reported range of 8–15 percentage points between low-end and high-end studies. That is a useful sanity check: headline numbers from any single paper should be read with caution, and meta-level comparisons like Fig. 3 are probably more honest than chasing the single best score in the literature.

| Application area | Representative methods | Acc / AUC |
|---|---|---|
| Oncology imaging & genomics | Deep CNNs, autoencoders, ensemble ML | 88–96% |
| Drug discovery & repurposing | Generative AI, graph NN, federated ML | 70–89% |
| Cardiovascular & metabolic | RNN/LSTM, wearable + DL, ensembles | 85–97% |
| Neurology & stroke | Multi-omics integration, autoencoders | 78–93% |
| Public-health surveillance | Time-series ML, transformer models | 80–91% |
| Mental-health triage | NLP, scoping-review classifiers | 76–91% |
| Pancreatic / breast imaging | Deep CNNs with feature selection | 88–98% |

Table I. Summary of application areas, representative methods and reported performance ranges across the reviewed studies.

## 6. DISCUSSION

Reading the reviewed literature in one sitting makes a few patterns hard to miss. The first is that the field has moved past the proof-of-concept stage. We are no longer asking whether an ML model can match a human reader on a fixed benchmark; we are asking whether a model can be deployed safely, audited fairly and updated without breaking clinical workflows. That is a different and much harder question.

The second pattern is that interpretability is becoming the gating factor. Several reviewed studies report excellent accuracy but acknowledge, sometimes only in passing, that clinicians remain reluctant to act on outputs they cannot explain. Counterfactual XAI (Alam et al., 2026) and knowledge-graph reasoning (Uddin et al., 2025) both look promising, but neither is yet a default. Until they are, the gap between research-grade and clinic-grade AI will stay wider than headline numbers suggest.

The third pattern is structural. Federated learning, privacy-preserving computation and explainability all individually work; the challenge is integrating them. A federated model that is also explainable, also fair across demographics, and also fast enough to live inside a clinical decision-support tool is rarer than it should be. We see this as the central engineering problem of the next few years — not the algorithms in isolation, but the system around them. Table II summarizes the most consistently reported challenges and the mitigations the literature has converged on.

| Challenge | Why it matters | Promising mitigations |
|---|---|---|
| Limited external validation | Models that look excellent on one cohort fail in another hospital. | Multi-site federated training; benchmark suites; pre-registered validation. |
| Black-box decisions | Clinicians and regulators need to understand why a recommendation was made. | SHAP/LIME, counterfactual XAI, knowledge-graph reasoning (Alam et al., 2026; Uddin et al., 2025). |
| Privacy of patient data | Genomic and EHR data are highly sensitive and legally protected. | Federated learning, homomorphic encryption, synthetic data (Orthi et al., 2025). |
| Bias and fairness | Under-represented groups receive less accurate predictions. | Fairness-aware training; demographic audits; human-in-the-loop review. |
| Regulatory uncertainty | Approval pathways for adaptive AI are still maturing. | Model cards; lifecycle monitoring; alignment with FDA/EMA SaMD guidance. |
| Workflow integration | Even an accurate model fails if clinicians cannot fit it into their day. | Embedded decision support; QA-driven testing; digital twins for safe rollout (Rahman et al., 2025b). |

Table II. Persistent challenges in AI-enabled precision medicine, why each one matters in practice, and the most frequently cited mitigations across the reviewed literature.

A final note on equity. Several reviewed studies call out, explicitly, that AI in medicine risks reproducing the inequalities of the data it learns from. Rural and low-resource settings remain under-represented (Moniruzzaman et al., 2025), and demographic minorities are under-represented in genomic reference panels. Technical fixes — reweighting, federated training across diverse sites, help, but they are not a substitute for deliberate, inclusive data collection. We mention this as a research priority, not a complaint.

## 7. CONCLUSION AND FUTURE WORK

This review has tried to do three things at once: synthesize a fragmented literature on AI in bioinformatics and precision medicine, propose a layered framework that captures the shared architecture across that literature, and put a numerical floor and ceiling on what current models actually deliver. The picture that emerges is cautiously optimistic. Reported accuracies above 90% are no longer rare in oncology imaging and cardiovascular monitoring; multi-omics integration is producing biologically plausible biomarker candidates (Manik, 2023; Manik et al., 2022; Manik et al., 2021a); and federated, privacy-preserving designs are beginning to address the sharing problem that has held the field back for years (Orthi et al., 2025).

Real adoption will depend on engineering, not just algorithms. Better external validation, first-class explainability, fairness audits and tighter integration with clinician workflows are the levers that matter most over the next research cycle. We expect to see more digital-twin-based pre-deployment testing, more counterfactual reasoning, more cross-hospital benchmarks, and we hope more deliberate inclusion of under-represented populations in training and evaluation.

In closing, AI is not going to replace the clinical encounter. The reviewed evidence does not support that claim, and we do not believe it should. What AI can do, and is increasingly doing, is hand the clinician a sharper, faster, more personalized picture of the patient in front of them. That alone is a meaningful change and it is the change worth building for.

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