



Artificial Intelligence in Medical Laboratory Diagnostics: Current Applications, Challenges and Future Directions

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ABSTRACT :

Artificial intelligence (AI), including machine learning and deep learning, is increasingly being incorporated into medical laboratory diagnostics. Clinical laboratories generate large volumes of structured numerical, categorical, image-based, molecular and longitudinal data, creating an environment in which AI can support pattern recognition, classification, prediction, workflow optimization and decision support. This narrative review summarizes current applications of AI across the laboratory diagnostic pathway, with emphasis on pre-analytical processes, clinical chemistry, hematology, microbiology, molecular diagnostics, transfusion medicine and digital pathology. AI can assist with specimen quality assessment, result validation, cell and microorganism classification, image interpretation, antimicrobial susceptibility prediction, molecular data analysis and identification of clinically relevant patterns across multimodal datasets. However, high reported model performance does not automatically establish clinical usefulness. Major barriers include limited external and multicentre validation, dataset shift, class imbalance, hidden bias, poor interoperability, cybersecurity and privacy concerns, explainability limitations, workflow disruption, regulatory uncertainty, costs and the need for continuing human oversight. Recent reporting and governance frameworks, including TRIPOD+AI, STARD-AI and FUTURE-AI, emphasize transparent reporting, rigorous evaluation, fairness, traceability, usability, robustness and explainability. The future of laboratory AI is therefore likely to depend less on isolated high-performing algorithms and more on validated, interoperable and clinically integrated systems that augment laboratory professionals rather than replace them. Multidisciplinary collaboration among laboratory scientists, clinicians, pathologists, data scientists, engineers, regulators and patients will be essential for safe and equitable implementation.

Keywords: artificial intelligence; machine learning; deep learning; medical laboratory digital pathology; biomarkers;

1. Introduction

Medical laboratory diagnostics are central to contemporary healthcare, supporting screening, diagnosis, prognosis, treatment selection and monitoring. Modern laboratories increasingly use automated analytical platforms, laboratory information systems, digital microscopy, high-throughput sequencing and other technologies that generate large and complex datasets. The increasing volume and heterogeneity of these data have created opportunities for computational methods capable of identifying patterns that may be difficult to detect using conventional rule-based approaches.

Artificial intelligence refers broadly to computational methods that perform tasks associated with human intelligence, while machine learning (ML) refers to methods that learn relationships from data. Deep learning (DL), a subset of ML, uses multilayer neural networks and is particularly important for image, signal and high-dimensional data analysis. In laboratory medicine, AI is being explored across the complete testing pathway rather than only at the analytical stage. A 2024 review described applications of AI across different stages of clinical testing and highlighted the continuing gap between research models and clinically approved tools (Li et al., 2024).

The laboratory environment is particularly suitable for AI because much laboratory information is structured, high-volume and generated under standardized procedures. Earlier reviews emphasized the importance of laboratory professionals in ensuring that laboratory data are appropriately represented and that AI-supported diagnostics are safe and clinically meaningful (Lippi & Mattiuzzi, 2024). More recent work has broadened this perspective to include workflow automation, digital pathology, hematology, microbiology, pre-analytical quality, genomics and multimodal decision support.

This review examines the current role of AI in medical laboratory diagnostics, identifies the principal technical and clinical barriers to implementation, and discusses future directions for responsible adoption. The emphasis is on AI as a decision-support and workflow-enhancing technology, with final clinical responsibility remaining with appropriately trained healthcare professionals.

2. Scope and Review Approach

This article is a narrative review based on literature identified through PubMed-indexed publications, major medical and laboratory journals, and authoritative guidance from organizations including the World Health Organization (WHO) and the U.S. Food and Drug Administration (FDA). Recent literature from 2020–2026 was emphasized, while seminal reporting and governance frameworks were retained where necessary. The search concepts included artificial intelligence, machine learning, deep learning, clinical laboratory, laboratory medicine, medical diagnostics, digital pathology,

hematology, microbiology, clinical chemistry, molecular diagnostics, antimicrobial susceptibility testing, pre-analytical errors, validation, explainability and regulation. Because this is a narrative review rather than a systematic review, no pooled effect estimate or formal risk-of-bias synthesis was attempted.

3. AI Technologies Relevant to Laboratory Diagnostics

Technology	Typical laboratory use	Key consideration
Machine learning	Classification, prediction, anomaly detection, autoverification	Requires representative training and validation data
Deep learning	Microscopy, digital pathology, image and signal analysis	Data intensive; risk of overfitting and dataset shift
Computer vision	Cell, parasite, microorganism and tissue-image analysis	Image quality and annotation strongly affect performance
Natural language processing	Report extraction, coding, literature and clinical-text support	Language ambiguity and hallucination risk
Generative AI / multimodal models	Report assistance, information synthesis, multimodal decision support	Output verification, privacy and governance are essential
Hybrid/rule-based AI	Combining laboratory rules with learned models	Can improve transparency but requires careful rule maintenance

4. Applications Across the Laboratory Diagnostic Pathway

4.1 Pre-analytical Phase

The pre-analytical phase includes test ordering, patient preparation, specimen collection, identification, transport, centrifugation and processing. Errors at this stage can compromise downstream analytical quality. AI may assist in identifying patterns associated with unsuitable specimens, hemolysis, insufficient volume, clotting, delayed transport, incorrect tube selection or other pre-analytical problems. AI applications in the pre-analytical phase include automated recognition and prediction of specimen-related errors (Lippi et al., 2024).

AI-based systems may also combine specimen metadata with historical laboratory information to predict the likelihood of recollection or identify unusual pre-analytical patterns. Such systems should be validated against local collection practices because specimen types, transport systems, instrumentation and error frequencies vary among institutions.

4.2 Clinical Chemistry

Clinical chemistry generates large volumes of numerical results that can be interpreted individually or as patterns. AI can support autoverification, detection of analytical anomalies, prediction of clinically relevant combinations of laboratory results, reference-interval research and identification of patients requiring further investigation. Machine learning can potentially integrate repeated measurements rather than treating each laboratory result as an isolated observation.

The principal opportunity is therefore not simply faster result generation, but more contextual interpretation of laboratory data. However, models trained on one population may perform differently after changes in analyzers, reagents, calibration, patient mix or clinical practice. Continuous performance monitoring is consequently important.

4.3 Hematology

Hematology is one of the most active areas of laboratory AI because blood and bone marrow specimens generate image-based, numerical and flow-cytometric data. AI-assisted microscopy can support identification and classification of leukocytes, erythrocyte abnormalities, platelet morphology and abnormal cells. In digital hematology, convolutional neural networks and other deep-learning methods can automate image segmentation and classification. Recent reviews describe applications in peripheral blood and bone marrow morphology, flow cytometry, cytogenetics, genomics and hemostasis (Osman, 2026; Artificial Intelligence in Haematologic Diagnostics, 2025).

In flow cytometry, AI may assist with automated gating and recognition of abnormal populations. In genomics and cytogenetics, machine-learning approaches can support variant classification and pattern recognition. Nevertheless, hematologic diagnosis frequently requires integration of morphology, immunophenotype, cytogenetics and molecular findings; therefore, AI should be evaluated as part of the complete diagnostic pathway rather than as an isolated image classifier.

4.4 Microbiology and Antimicrobial Susceptibility Testing

Microbiology presents opportunities for AI in organism recognition, microscopy, colony classification, identification of pathogens, antimicrobial susceptibility testing (AST), resistance prediction and surveillance. Recent reviews describe the use of machine learning, random forests, support vector machines, convolutional neural networks and transformer-based approaches in bacterial diagnostics and antimicrobial susceptibility testing (Lee et al.,

2025; Alsulimani et al., 2024).

AI may be particularly valuable where rapid preliminary classification or resistance prediction could shorten the time to appropriate therapy. However, predictive performance can be strongly affected by local resistance epidemiology and the distribution of organisms and resistance mechanisms. Models should therefore be externally validated and monitored when resistance patterns change.

4.5 Molecular Diagnostics and Genomics

Molecular laboratories generate high-dimensional datasets from PCR, sequencing and other molecular platforms. AI can assist with sequence classification, variant interpretation, genomic pattern recognition, prediction of antimicrobial resistance and integration of molecular findings with clinical and laboratory variables. The value of AI in this setting lies in reducing the burden of high-dimensional interpretation while preserving analytical and clinical safeguards.

A major challenge is the distinction between computational prediction and clinical significance. A model can classify a sequence or variant accurately without proving that the resulting interpretation improves patient outcomes. Clinical validation and clear definitions of intended use are therefore essential.

4.6 Histopathology, Cytology and Digital Pathology

Digital pathology is among the most visible areas of clinical AI. Whole-slide images can be analyzed using computer vision and deep-learning methods for tissue segmentation, cell detection, classification, tumor detection, grading and quantitative biomarker assessment. A 2024 systematic review and meta-analysis reported high mean sensitivity and specificity across included AI diagnostic studies, but also found substantial heterogeneity and high or unclear risk of bias in most studies, emphasizing that reported accuracy does not by itself establish clinical readiness (McGenity et al., 2024).

AI is also being studied in cytology and computational pathology for screening and diagnostic assistance. More recent work has expanded toward generative AI and multimodal systems. However, pathology AI requires attention to staining variation, scanner differences, tissue preparation, annotation quality and population differences. Human review remains important, especially for ambiguous or clinically consequential findings.

The rapid development of generative AI introduces additional possibilities for pathology, including report assistance, image-text interaction and multimodal information synthesis. A 2025 review of generative AI in anatomic pathology noted potential applications in diagnostic workflow and research while also highlighting the need for careful evaluation.

4.7 Transfusion Medicine

Potential laboratory applications of AI in transfusion medicine include donor and component-demand forecasting, blood inventory optimization, identification of unusual compatibility patterns, antibody investigation support and adverse-event surveillance. These applications are particularly dependent on high-quality historical data and strong governance because incorrect recommendations can have serious consequences. AI should therefore remain within validated decision-support boundaries and established transfusion policies.

4.8 Laboratory Quality Management and Autoverification

AI can support quality management by identifying unusual analytical patterns, predicting instrument or process failures, prioritizing quality-control investigations and improving autoverification. Unlike simple fixed rules, ML models can learn from historical distributions and combinations of variables. However, AI-based quality systems should not obscure established quality-control requirements; rather, they should complement them. Any automated release or rejection rule should have defined thresholds, documented validation and a safe manual override mechanism.

5. Potential Benefits of AI in Laboratory Diagnostics

Potential benefit	Laboratory example	Important qualification
Speed	Automated image screening or result prioritization	Speed should not reduce verification quality
Consistency	Standardized image classification	Performance depends on data and preprocessing
Scalability	Analysis of large datasets	Infrastructure and interoperability are required
Pattern recognition	Multivariable laboratory-result interpretation	Association does not prove causation
Early detection	Risk prediction from longitudinal laboratory data	Prospective clinical benefit must be demonstrated
Workflow support	Autoverification and triage	Human oversight and escalation pathways are needed
Quality improvement	Detection of abnormal analytical patterns	Local validation is essential

6. Major Challenges and Limitations

6.1 Data Quality and Representativeness

AI systems learn from the data supplied to them. Missing values, inconsistent labels, measurement differences, class imbalance and changes in patient populations can produce misleading models. Laboratory data may also contain institution-specific patterns that are not generalizable. External validation on independent populations is therefore a central requirement before clinical deployment.

6.2 Dataset Shift and Model Drift

Laboratory systems change over time. New analyzers, reagents, reference intervals, collection procedures, disease prevalence and clinical guidelines can alter the relationship between inputs and outcomes. A model that performs well at deployment may deteriorate later. AI governance should therefore include post-deployment monitoring, recalibration and predefined criteria for suspension or revalidation.

6.3 Explainability and the 'Black Box' Problem

Some deep-learning models provide limited intuitive explanations for individual predictions. Explainability is particularly important when an AI output influences a high-stakes laboratory decision. However, an explanation method should not be assumed to prove that the underlying model is clinically correct. Explainability should be combined with robust validation, calibration, uncertainty assessment and appropriate human oversight.

The World Health Organization emphasizes transparency, explainability and intelligibility as part of responsible AI in health (World Health Organization, 2021).

6.4 Bias and Health Equity

Bias can arise from under-representation of demographic groups, differences in disease prevalence, unequal access to diagnostic services or systematic differences in specimen collection and laboratory procedures. A model may therefore perform differently across populations. Fairness assessment should be planned during development and repeated after deployment rather than treated as a one-time exercise.

6.5 Privacy, Cybersecurity and Data Governance

Laboratory datasets can contain identifiable and clinically sensitive information. AI development and deployment require appropriate de-identification, access controls, secure storage, audit trails and governance of data sharing. Generative and multimodal AI create additional concerns because sensitive information may be transmitted to external systems or incorporated into model workflows. WHO guidance emphasizes privacy, human rights, accountability and appropriate governance (World Health Organization, 2021, 2025).

6.6 Regulatory and Legal Issues

AI-enabled medical technologies are increasingly subject to regulatory oversight. The U.S. Food and Drug Administration maintains an AI-enabled medical-device list and describes evaluation within existing medical-device regulatory pathways, with attention to the product lifecycle including development, validation, deployment, monitoring and modification (U.S. Food and Drug Administration, 2026).

Regulatory requirements vary across jurisdictions. Laboratories adopting AI should define intended use, analytical and clinical performance requirements, responsibility for errors, change-control procedures and post-market or post-deployment monitoring before implementation.

6.7 Interoperability and Workflow Integration

An accurate algorithm can still fail to deliver clinical value if it cannot communicate reliably with the laboratory information system, analyzer, digital pathology platform or electronic health record. Workflow design should specify who receives the AI output, when it is displayed, what action is expected and how disagreement between the AI and laboratory professional is resolved.

6.8 Cost, Skills and Infrastructure

AI implementation may require digital infrastructure, high-quality data storage, network capacity, validated software, cybersecurity controls and trained personnel. Laboratory professionals therefore need AI literacy, while data scientists need understanding of laboratory processes, specimen characteristics, quality systems and clinical consequences. Interdisciplinary teams are essential.

7. Validation and Clinical Implementation

AI validation should progress beyond retrospective internal testing. A practical pathway includes technical verification, internal validation, external validation, prospective clinical evaluation, workflow assessment and post-deployment monitoring. Evaluation should include discrimination, calibration, clinically relevant thresholds, subgroup performance, failure modes and human factors where appropriate.

The DECIDE-AI guideline emphasizes early clinical evaluation of AI decision-support systems because preclinical or in-silico performance does not necessarily translate into clinical benefit (Vasey et al., 2022). For prediction models, TRIPOD+AI provides updated reporting recommendations applicable to regression and machine-learning models (Collins et al., 2024). For AI-based diagnostic accuracy studies, STARD-AI provides reporting considerations specific to AI-centered diagnostic studies (STARD-AI Steering Group, 2025).

Implementation should also consider the total AI lifecycle. FUTURE-AI provides an international consensus framework organized around fairness, universality, traceability, usability, robustness and explainability, with recommendations spanning development, validation, regulation, deployment and monitoring (Lekadir et al., 2025).

8. Role of Medical Laboratory Professionals

AI is likely to change the role of laboratory professionals rather than eliminate the need for them. Laboratory scientists, pathologists and other specialists possess domain knowledge needed to define appropriate use cases, curate and label data, recognize pre-analytical and analytical artifacts, validate models, assess clinically meaningful errors and monitor performance. They can also determine when an AI recommendation is inconsistent with specimen quality or the broader clinical context.

Training programs in medical laboratory science should increasingly include basic concepts of AI, data quality, statistics, model performance, validation, bias, privacy and ethical use. AI literacy should be viewed as an extension of laboratory quality competence rather than solely as a computer-science skill.

9. Generative AI and Multimodal Diagnostics

Generative AI and large multimodal models represent a newer phase of AI development. These systems can process or generate text, images and other data modalities and may eventually support integrated interpretation of laboratory reports, pathology images, clinical information and molecular findings. WHO's 2025 guidance on large multimodal models recognizes their potential use in healthcare while emphasizing governance and the uncertainty surrounding their broad clinical capabilities (World Health Organization, 2025).

In laboratory medicine, a cautious approach is warranted. Generative models can produce plausible but incorrect statements, and their outputs may be difficult to reproduce. They should therefore be used within defined tasks, with source verification, restricted access to sensitive data, human review and auditability. Their use for direct diagnostic decision-making should require substantially stronger evidence than their use for administrative or educational assistance.

10. Future Directions

Future laboratory AI research is likely to move from isolated single-task algorithms toward integrated systems capable of combining laboratory values, images, molecular data and longitudinal clinical information. Several developments are particularly important:

- **Multimodal AI:** Integration of numerical, image, molecular and clinical-text data.
- **Federated and privacy-preserving learning:** Collaborative model development without unnecessary transfer of identifiable data.
- **Explainable and uncertainty-aware AI:** Systems that communicate confidence, limitations and reasons for predictions.
- **Continuous monitoring:** Detection of model drift and performance changes after deployment.
- **Human-AI collaboration:** Workflows designed around complementary strengths rather than replacement of professionals.
- **Standardized evaluation:** Greater use of reporting and evaluation frameworks such as TRIPOD+AI, STARD-AI, DECIDE-AI and FUTURE-AI.
- **Resource-sensitive AI:** Efficient systems suitable for laboratories with limited infrastructure and specialist availability.

11. Proposed Framework for Responsible AI Adoption in Medical Laboratories

1. Define a specific clinical or laboratory problem and intended use.
2. Assess data quality, representativeness, privacy and governance requirements.
3. Develop or select the AI model using appropriate training procedures.
4. Perform analytical verification and internal validation.
5. Perform independent external validation across relevant instruments, sites and populations.
6. Conduct prospective clinical and workflow evaluation.
7. Integrate the model with laboratory information and quality systems.
8. Train laboratory personnel and define escalation and override procedures.
9. Monitor performance, bias, safety, cybersecurity and model drift continuously.
10. Revalidate or retire the model when performance, data sources or intended use changes.

12. Discussion

The evidence base indicates that AI has moved beyond a theoretical concept in laboratory medicine and is being investigated or implemented across multiple diagnostic domains. The strongest current opportunities are associated with repetitive, data-rich and pattern-recognition tasks such as image

classification, workflow triage, quality monitoring and interpretation of high-dimensional datasets. Recent literature in hematology, microbiology and digital pathology demonstrates rapid technical development, while surveys of laboratory medicine indicate that real-world adoption remains more limited than the volume of research might suggest (Osman, 2026; Lee et al., 2025; Cadamuro et al., 2025).

The central challenge is therefore translational. A model with high sensitivity or accuracy under controlled research conditions may not improve patient care when introduced into a heterogeneous laboratory environment. Diagnostic accuracy, clinical utility, workflow effects, safety, fairness, usability and sustainability all need to be considered. The 2024 digital-pathology meta-analysis illustrates this distinction: high pooled diagnostic performance coexisted with substantial concerns regarding risk of bias and applicability (McGenity et al., 2024).

Responsible implementation also requires recognition that laboratory professionals remain accountable for specimen quality, analytical validity and clinical interpretation within their professional scope. AI should be positioned as a validated component of the laboratory system, not as an autonomous authority. WHO guidance similarly places human autonomy, safety, transparency, accountability and equity at the center of AI governance (World Health Organization, 2021).

13. Limitations of This Review

This narrative review was designed to provide a broad overview rather than a systematic evidence synthesis. The literature is heterogeneous with respect to algorithms, datasets, reference standards, endpoints and clinical settings, making direct comparison of reported performance difficult. Many published studies remain retrospective, single-center or focused on technical performance. Rapid changes in AI technology also mean that specific tools and regulatory positions may change after publication. Future systematic reviews should evaluate diagnostic accuracy, clinical impact and implementation outcomes separately for each laboratory domain.

14. Conclusion

Artificial intelligence is becoming an important component of the evolving medical laboratory ecosystem. Its applications extend from pre-analytical quality and workflow automation to hematology, microbiology, molecular diagnostics, clinical chemistry and digital pathology. The technology offers substantial potential for improving consistency, scalability and pattern recognition, but evidence of technical performance should not be equated with proof of clinical benefit. The principal priorities for the next phase of laboratory AI are external validation, transparent reporting, bias assessment, interoperability, cybersecurity, lifecycle monitoring and prospective evaluation of clinical utility. Successful adoption will require collaboration between laboratory professionals and AI specialists and should preserve meaningful human oversight. The future of AI in laboratory diagnostics is therefore most appropriately framed as augmented laboratory medicine: computational systems supporting expert professionals within validated, safe and accountable workflows.

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