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Role of Machine Learning in Diagnostic Pathology and Laboratory Medicine: A Narrative Review

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Abstract

Objective: Disease screening and diagnosis provide common issues for pathology and clinical laboratory medicine. These challenges include growing workloads, staffing shortages, diagnostic variability, and error susceptibility across the entire testing process. Although machine learning (ML) has been extensively used in image analysis and laboratory automation, studies that have already been written frequently focus on these areas independently, which prevents a comprehensive understanding of how ML affects diagnostic practice. This narrative review's goal is to compile the most recent data on machine learning applications in pathology and clinical laboratory medicine, with a focus on workflow efficiency, diagnostic performance, and the identification of laboratory errors throughout the entire testing process. Methods: Using predetermined search terms associated with machine learning, pathology, clinical laboratory medicine, and diagnostic quality, we performed a narrative review of studies indexed in PubMed, Scopus, Web of Science, and IEEE Xplore. Four domains diagnostic image analysis, biomarker and outcome prediction, workflow automation, and laboratory error detection are combined to create ML applications. Applying deep learning (DL) models to whole-slide images (WSIs) in pathology has produced results that are on par with pathologists in controlled environments. By utilizing multianalyte and temporal patterns, ML-based systems in clinical laboratory medicine improve automated verification, patient-based quality control (QC), and error identification, frequently exceeding conventional rule-based techniques. Prognostic modeling and decision support are further enhanced by new multimodal frameworks that incorporate laboratories, clinical, and histopathologic data. Conclusions: This study emphasizes the potential and constraints of machine learning (ML) in diagnostic medicine, stressing the necessity of interpretability, generalizability, and regulatory supervision to enable clinically significant, integrated implementation.

Keywords: *Machine Learning, Diagnostic Pathology, Laboratory Medicine.*

Introduction

The term "machine learning" (ML) describes computational techniques that allow algorithms to enhance their performance on certain tasks without explicit programming by learning patterns from data. ML encompasses a wide range of techniques, from contemporary deep learning (DL) and neural network topologies to traditional statistical models like logistic regression and decision trees. The task's complexity, the application's therapeutic context, and

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the data's structure all influence which approach is best (Rajagopal et al., 2024).

By facilitating the automated, precise, and scalable interpretation of intricate medical pictures and laboratory data, machine learning has started to revolutionize the discipline of pathology in recent years. Applications including prognostic risk modeling, diagnostic decision support, and histopathologic picture categorization are supported by its paradigm-shifting incorporation into diagnostic workflows. It has been demonstrated that these instruments increase diagnostic accuracy, boost consistency, shorten turnaround times, and lessen manual labor. Early machine learning applications in pathology relied on manually created feature extraction under the direction of subject-matter knowledge, which restricted scalability and generalizability. However, the development of DL has made it possible to learn categorized features directly from raw image data, significantly enhancing performance in challenging tasks including metastasis identification, tumor classification, and margin detection (Andersen et al., 2024; Rathore et al., 2020).

Pathology and clinical laboratory medicine still encounter frequent and enduring difficulties during disease screening and diagnosis, despite advancements in diagnostic tools. These factors, which put pressure on diagnostic accuracy, consistency, and turnaround time, include staff shortages, continually rising test volumes, and increasing case complexity. Manually interpreting complicated histologic patterns in pathology takes a lot of time and is prone to interobserver variability, especially for lesions that are borderline or heterogeneous. While conventional rule-based quality control (QC) systems might not be sensitive to subtle or multivariate error patterns, high-throughput testing across the whole testing process in clinical laboratories creates several opportunities for pre-analytical, analytical, and post-analytical errors.

Applications of machine learning (ML) in ordinary clinical laboratory medicine or diagnostic pathology have been the subject of several recent reviews, frequently with an emphasis on image analysis, automation, or biomarker identification (Litjens et al., 2017).

The comprehensive scope and outcome-oriented attitude of the current review set it apart. In the context of the entire testing process, we jointly investigate machine learning applications in pathology and clinical laboratory medicine, focusing on operational reliability, quality assurance, error detection, and diagnostic performance. This study also emphasizes new multimodal learning strategies that integrate laboratory, clinical, and histopathologic data, as well as real-world issues with interpretability, generalizability, and regulatory application. This narrative review offers a coherent and practice-relevant synthesis that enhances the body of current literature and illustrates the changing role of machine learning in diagnostic medicine by crossing traditionally divided diagnostic fields.

Methods

To find pertinent research on machine learning applications in pathology and clinical laboratory medicine, a systematic literature search was carried out on PubMed, Scopus, Web of Science, and IEEE Xplore. representing the current advancement and clinical application of ML and DL techniques.

High-quality reviews, multicenter studies, and original research articles written in English that discussed machine learning applications related to laboratory workflow, diagnostic interpretation, or quality assurance throughout the entire testing process were included. Studies with no clinical or operational significance and conference abstracts without peer-reviewed full publications were not included.

After screening articles for relevancy based on their title and abstract, the whole text was reviewed. With a focus on methodological diversity, clinical relevance, and practical implementation considerations, studies representing both established and new applications spanning pathological image analysis and clinical laboratory operations were chosen.

ML Applications in clinical laboratory medicine and pathology

While the goals of ML applications in pathology and clinical laboratory medicine are similar improving diagnostic quality, efficiency, and accuracy they differ in terms of data structure, workflow, and clinical contact points. While laboratory medicine applications rely on high-frequency, structured numerical data produced throughout the whole testing process, pathology applications primarily concentrate on image-based interpretation and spatial pattern recognition. Despite these distinctions, ML increasingly offers chances for integration, feedback, and coordinated decision support across historically divided areas, and both fields function inside interconnected diagnostic procedures (Esteva et al., 2019).

Diagnosis and image analysis

Image-based diagnosis is one of the main uses of machine learning in pathology. When it comes to tumor diagnosis, grading, and metastasis identification in a variety of cancers, such as breast, prostate, lung, and colorectal malignancies, machine learning models have performed on par with or better than human pathologists in controlled settings (Coudray et al., 2018).

The exceptional diagnostic potential of machine learning in histopathologic image processing has been shown by extensive research. Without the need for manual data curation, ML models were able to detect prostate cancer and breast cancer metastases with area under the curve (AUC) values of 0.991 and 0.966, respectively, in one study including over 44,000 WSIs from over 15,000 patients. For the detection of metastatic lymph nodes, another multicenter research found 97.1% accuracy, 93.5% positive predictive value (PPV), and 98.0% negative predictive value (NPV). When taken as a whole, these findings highlight the exceptional diagnostic accuracy and scalability of machine learning in well-annotated, image-based cancer diagnostics (Pantanowitz et al., 2014).

Prognostic prediction and biomarkers

ML, especially DL, has revolutionized biomarker identification and prognostic prediction in computational pathology beyond picture classification. According to recent research, DL can extract clinically significant characteristics from routine hematoxylin and eosin (H&E) stained WSIs. This allows for the prediction of molecular biomarkers and patient outcomes, supporting individualized treatment choices. Morphological patterns that are correlated with genetic alterations, protein expression, and survival outcomes include tissue texture, spatial

architecture, tumor microenvironment, and stromal organization, according to DL models (Rabbani et al., 2022; Yu et al., 2021).

In terms of prognostic modeling, DL and multimodal models that incorporate genetic and histologic data have demonstrated potential in predicting therapy response, overall survival, and disease recurrence. Multi-omic data integration improves prognostic accuracy and sheds light on biologically significant histologic correlations, including vascular proliferation, stromal remodeling, and tumor-infiltrating lymphocytes (Jariyapan et al., 2025).

Integrating laboratory, clinical, and histopathological data is known as multimodal machine learning

Multimodal machine learning (ML) is an emerging field in artificial intelligence (AI) that combines complementary data types to enhance diagnostic and prognostic performance, although most of the existing work approaches histopathologic image analysis and laboratory data modeling as distinct domains. When compared to unimodal techniques, models that simultaneously analyze WSIs with genetic, laboratory, and clinical factors have shown improved predictive accuracy in oncology and precision medicine. The advantages of each modality are combined in multimodal frameworks: while laboratory biomarkers and genomic data offer quantitative and molecular context reflecting tumor load, biology, and systemic response, WSIs capture spatial architecture, tumor microenvironment, and morphologic heterogeneity. For instance, DL models that incorporate genomic changes or circulating tumor markers with histopathologic features have demonstrated better performance than image-only or molecular-only models in predicting survival, recurrence risk, and treatment response (Echle et al., 2021; Acs, 2020).

Crucially, this paradigm organically connects clinical laboratory medicine with pathology. Tumor markers, inflammatory indices, metabolic profiles, and longitudinal chemistry trends are examples of laboratory data that are high-frequency, organized assessments that supplement static tissue appearance. Context-aware modeling of disease progression is made possible by integrating various data streams, which capture the temporal and spatial aspects of pathology. As multimodal learning frameworks develop, they provide a unifying strategy that unifies laboratory medicine and image-based diagnostics, enabling more precise risk assessment, individualized treatment planning, and outcome-focused assessment of diagnostic quality.

Efficiency and automation of workflow

ML-driven decision support systems have also improved process automation in clinical laboratory medicine. For instance, in high-throughput testing environments, such as mass spectrometry procedures, ML-based automated verification techniques have been developed to assess analytical quality and identify undesirable results. These technologies have proven to facilitate scalable laboratory operations while preserving analytical quality by detecting analytically incorrect data with high sensitivity and lowering the burden of manual review. These methods show how ML plays a common role in streamlining diagnostic workflows across disciplines, much like slide triage and case prioritizing in pathology (Hu Y et al., 2021;

Campanella et al., 2019).

ML for identifying mistakes in the lab

One of the most developed and practically useful uses of AI in clinical laboratory medicine is the detection of laboratory errors using machine learning (ML). These techniques are intended to monitor the integrity of the entire testing process by detecting aberrant patterns suggestive of pre-analytical, analytical, or post-analytical mistakes, in contrast to diagnostic prediction tasks that concentrate on illness classification. Together, recent research shows that machine learning (ML) has advantages over conventional rule-based systems, but it also raises significant issues with implementation, generalizability, and interpretability (Bulten et al., 2020; van, 2021).

The results of laboratory tests are used to guide diagnosis, prognosis, and treatment in the great majority of medical decisions. Therefore, there can be significant clinical and operational repercussions from laboratory errors, which are defined as any flaw that arises at any stage of the entire testing process and jeopardizes result integrity. Inappropriate test ordering and stewardship, issues with specimen collection and transportation, inaccurate analytical measurements, and post-analytical errors during auto-verification, reporting, or abnormal result flagging are all examples of laboratory errors that can happen at any point during the testing process (Wang et al., 2025).

Recommendations:

These limitations highlight the necessity of uniform evaluation frameworks, enhanced interpretability, and multicenter validation. Annotation and data accessibility continue to be some of the most basic obstacles. Large, carefully chosen data sets are necessary for ML algorithms to work dependably. It takes a lot of time and money for skilled pathologists to manually annotate such datasets in pathology, especially for WSI analysis. Data labeling is further complicated by the intricacy of histopathologic interpretation, variations in diagnostic criteria, and variances across observers. Furthermore, high-resolution WSIs have significant processing and storage needs, which frequently call for sophisticated image management systems, data servers, and specialized computational equipment. Accessibility may be hampered by these resource requirements, particularly for smaller organizations or research teams without specialized bioinformatics support.

The generalizability of the paradigm is another significant issue. When tested on data from the same institution or under comparable imaging conditions, machine learning models frequently perform well; nevertheless, their accuracy may deteriorate when applied to external datasets. Performance drift among sites can be caused by variations in staining procedures, imaging technology, scanning resolutions, and patient demographics. The necessity of multicenter datasets, cross-platform validation, and standardized data formats to enhance model transferability is highlighted by this lack of resilience.

Clinical adoption depends heavily on user trust and interpretability. Many pathologists' express skepticism toward algorithmic recommendations, citing concerns about reliability, potential job displacement, loss of interpretive autonomy, and increased documentation burden. To address

these issues, user-centered design, explainable AI (XAI) frameworks, and transparent validation studies are essential for fostering confidence and ensuring that ML systems augment rather than replace human expertise.

Finally, there are new obstacles to the practical application of machine learning in pathology and laboratory medicine due to ethical and regulatory issues. Adaptive learning algorithms that continuously change with data input were not intended for the current regulatory frameworks. The U.S. Food and Drug Administration oversees the regulation of various AI-enabled diagnostic tools in the country as Software as a Medical Device (SaMD), which calls for lifecycle-based risk management and premarket assessment of analytical validity and clinical performance. AI-based SaMD may continue to develop after deployment, in contrast to conventional static software or laboratory assays. This could lead to regulatory issues with model upgrades, performance monitoring, and version control.

Predetermined Change Control Plans (PCCPs), a regulatory concept that allows specified algorithm updates under regulatory supervision, have been established to solve these issues. PCCPs seek to strike a balance between innovation and patient safety throughout the AI lifecycle by outlining permissible model alterations and related validation techniques beforehand. However, concurrent attention to ethical issues including algorithmic bias, data privacy, informed consent, and auditability are also necessary for the effective governance of AI systems.

Conclusion

ML has quickly become a revolutionary force in laboratory medicine and pathology. Through quality control and error detection, it facilitates automated picture interpretation, increases diagnostic accuracy, aids prognostic modeling, and boosts operational effectiveness. ML offers novel capabilities that go well beyond conventional laboratory techniques, such as preanalytical mistake identification, analytical performance monitoring, and post-analytical interpretation. However, issues with data quality, interpretability, generalizability, workflow integration, and regulatory oversight continue to limit machine learning's full therapeutic potential. Establishing strong data standards, open-access repositories, and validation procedures will require cooperation between labs, data scientists, physicians, and regulatory bodies to overcome these obstacles.

Despite these challenges, progress is nevertheless happening at a faster rate. Model transparency is increasing and reliance on big annotated datasets is decreasing thanks to developments in federated learning, self-supervised training, and model explainability. Like this, standardized interfaces and automation pipelines are making it increasingly possible to integrate ML into digital pathology and LIS. As these developments advance, machine learning (ML) has the potential to move from experimental use into a reliable, comprehensible, and essential part of diagnostic practice, improving the effectiveness and precision of contemporary laboratory medicine.

There are many restrictions on this narrative review. First, the review is not a systematic meta-analysis and may be prone to selection bias or the omission of pertinent research, even if a structured literature search technique was used. Second, freshly published approaches and applications might not be fully represented due to the quickly changing nature of machine

learning research. Third, direct comparison of reported outcomes is hampered by variations in study design, datasets, performance indicators, and validation techniques within the collected literature. Lastly, a lot of the studies that are quoted rely on retrospective or single-institution data, which may overstate performance and generalizability in the real world. Further limiting the generalizability of reported ML performance across institutions are variations in digital pathology platforms, instrumentation, and laboratory operations. When interpreting the results and conclusions, these limitations should be considered.

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