



Multi-Omics Approaches in Diagnostic Pathology: An Updated Review of Current Evidence

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How to Cite the Article:

Mehta, Aarav, et al. "Multi-Omics Approaches in Diagnostic Pathology: An Updated Review of Current Evidence." *Journal of Advanced Biological Sciences*, vol. 02, no. 02, December 2025, pp. 20-21. <https://doi.org/10.66590/jabs2025020204>

Abstract | Diagnostic pathology has undergone a remarkable transformation from a predominantly morphology-based discipline to an integrated field that combines histopathology, immunohistochemistry, molecular pathology, and computational biology. The emergence of multi-omics approaches—including genomics, transcriptomics, proteomics, metabolomics, epigenomics, and spatial omics—has enabled comprehensive characterization of disease biology by providing complementary molecular insights. These technologies have significantly advanced precision medicine by improving disease classification, facilitating early and accurate diagnosis, identifying clinically actionable biomarkers, predicting therapeutic response, and uncovering mechanisms of treatment resistance. Furthermore, the integration of multi-omics data through advanced computational and artificial intelligence-driven analytical methods has enhanced diagnostic accuracy and personalized clinical decision-making. This review provides a comprehensive overview of current multi-omics technologies, their applications in diagnostic pathology, computational integration strategies, clinical significance, existing challenges, and future directions for their implementation in precision diagnostics and personalized medicine.

Key Words Multi-omics, Diagnostic pathology, Genomics, Transcriptomics, Proteomics, Precision medicine, Molecular pathology

INTRODUCTION

Traditional pathology has relied primarily on microscopic examination of tissue morphology. While morphology remains fundamental, advances in molecular technologies have transformed pathology into a multidisciplinary field capable of characterizing diseases at multiple biological levels. Multi-omics integrates diverse molecular datasets to provide a comprehensive understanding of disease biology that cannot be achieved using a single analytical platform [1].

Major Multi-Omics Technologies

- Genomics: DNA mutations, copy number alterations, structural variants, and inherited susceptibility
- Transcriptomics: Gene-expression profiling, RNA sequencing, alternative splicing, and non-coding RNA analysis

- Proteomics: Protein expression, post-translational modifications, and signaling pathways
- Metabolomics: Small-molecule metabolites reflecting cellular metabolism
- Epigenomics: DNA methylation, histone modifications, and chromatin accessibility
- Spatial Omics: Molecular information preserved within tissue architecture [2]

Applications in Diagnostic Pathology

Multi-omics contributes to:

- Improved tumor classification
- Identification of diagnostic biomarkers
- Prognostic risk stratification
- Prediction of therapeutic response

- Detection of minimal residual disease
- Characterization of rare tumors
- Precision oncology and personalized treatment

Examples include molecular classification of brain tumors, integrated diagnosis of hematologic malignancies, molecular profiling of breast carcinoma, colorectal cancer, lung cancer, thyroid carcinoma, and soft tissue sarcomas [3-5].

Role of Artificial Intelligence

Artificial intelligence assists in [6]:

- Integrating heterogeneous omics datasets
- Predicting molecular alterations from histopathology
- Biomarker discovery
- Survival prediction
- Automated patient stratification
- Clinical decision support

Advantages

Advantage	Clinical Impact
Comprehensive disease characterization	Improved diagnostic accuracy
Biomarker discovery	Earlier diagnosis
Precision medicine	Personalized therapy
Molecular subclassification	Better prognostic assessment
Treatment prediction	Optimized patient management

Challenges

Major limitations include:

- High analytical cost
- Large data storage requirements
- Complex bioinformatic analysis
- Lack of standardized workflows
- Limited access in resource-constrained laboratories
- Data privacy and ethical considerations
- Need for multidisciplinary expertise

Future Perspectives

Emerging directions include:

- Single-cell multi-omics
- Spatial transcriptomics integrated with digital pathology

- AI-driven multi-modal diagnostics
- Cloud-based bioinformatics platforms
- Routine clinical implementation of integrated molecular pathology
- Real-time precision diagnostics using combined histological and molecular data

CONCLUSIONS

Multi-omics approaches are redefining diagnostic pathology by integrating genomic, transcriptomic, proteomic, metabolomic, and epigenomic information with conventional histopathology. These technologies are enhancing diagnostic precision, improving prognostic evaluation, and supporting individualized treatment decisions. Continued advances in artificial intelligence, computational pathology, and standardized clinical workflows are expected to make multi-omics an increasingly important component of routine pathology practice.

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