

Letter to Editor

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AI Based Molecular Diagnostics: Emerging Challenges in Standardization

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Abstract

Recent advances in artificial intelligence (AI) and machine learning have substantially expanded their application in molecular diagnostic technologies. AI-based tools are increasingly integrated into various stages of molecular testing workflows, including primer and probe design, interpretation of real-time and digital PCR data, and high-throughput analysis of next-generation sequencing (NGS) datasets. While these technologies offer significant advantages in terms of analytical speed, accuracy, and reduction of human error, their rapid implementation has outpaced the development of harmonized standards and regulatory frameworks.

This article provides a critical analysis of the emerging challenges associated with the standardization of AI-assisted molecular diagnostics. Current international standards for molecular testing have been primarily designed for conventional analytical methodologies and do not adequately address algorithm-driven decision-making processes. Key challenges discussed include data quality and governance, algorithm training and validation strategies, software version control, model adaptability, and the lack of standardized performance evaluation metrics tailored to AI-based systems. These limitations contribute to inter-laboratory variability, reduced reproducibility, and uncertainty in regulatory acceptance.

The regulatory and societal implications of deploying AI-based molecular diagnostics, particularly in food safety, agriculture, and public health, are also examined. Finally, the article emphasizes the urgent need for dedicated and harmonized standards that encompass analytical performance, algorithm lifecycle management, transparency, and quality assurance. Addressing these gaps is essential for ensuring result reliability, regulatory confidence, and the sustainable integration of AI-driven molecular diagnostic technologies.

Keywords Artificial Intelligence, Molecular Diagnostics, Standardization, Regulatory Frameworks, Algorithm Validation

Introduction

Recent advances in artificial intelligence (AI) and machine learning have significantly expanded their application in molecular diagnostic technologies. AI based tools are now increasingly used in various stages of molecular testing workflows, including primer and probe design, interpretation of real time PCR amplification curves, digital PCR data analysis, and the processing of high throughput

next generation sequencing (NGS) datasets [1,2]. While these technologies offer substantial advantages in terms of analytical speed, data interpretation, and reduction of human error, their rapid adoption has outpaced the development of appropriate standards and regulatory frameworks [1,3]. The absence of harmonized and transparent standards for AI assisted molecular diagnostics raises critical concerns regarding data reliability, regulatory ac-



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ceptance, and inter laboratory comparability [4].

1. The Role of Artificial Intelligence in Molecular Diagnostics

AI driven algorithms enable the identification of complex biological patterns that are often undetectable using conventional analytical approaches [1,5]. In molecular diagnostics, these systems rely heavily on large training datasets and advanced learning models to enhance sensitivity, specificity, and decision making accuracy [1,6]. However, algorithm performance is intrinsically dependent on data quality, training strategies, and laboratory specific conditions [1,2]. In the absence of standardized guidelines governing these parameters, variability in analytical outcomes between laboratories becomes inevitable, potentially undermining the robustness of diagnostic conclusions [4,7].

2. Limitations of Current Standard Frameworks

Existing international standards for molecular testing, including ISO/IEC 17025 and method validation guidelines, have been primarily developed for classical analytical methodologies [8]. These frameworks typically assume direct interpretation of experimental outputs without considering the integration of algorithmic decision making layers [8,9]. Consequently, critical aspects such as algorithm training procedures, data selection criteria, software version control, and model adaptability are not explicitly addressed [8,10]. This gap creates ambiguity in defining method scope, reproducibility, and traceability when AI based tools are incorporated into routine diagnostic practice [4,9].

3. Performance Evaluation and Result Reliability

A major challenge lies in the lack of standardized performance metrics tailored to AI assisted molecular diagnostics [1,4]. While analytical sensitivity, specificity, limit of detection, and precision are well established for conventional molecular assays, their translation to algorithm driven systems remains insufficiently defined [1,4]. For example, it is often unclear when algorithm retraining or software updates necessitate re validation of the diagnostic method [4,7]. Without clear criteria, the regulatory credibility of AI based molecular results may be compromised, particularly in contexts where diagnostic outcomes inform public health or food safety decisions [1,3,4].

4. Regulatory Implications and Public Trust

In critical areas such as food safety, agriculture, and public health, molecular diagnostic results play a pivotal role in regulatory decision making [2,3]. The deployment of

AI based diagnostic systems without standardized requirements for algorithm transparency, explainability, and result traceability may challenge both regulatory oversight and public confidence [5,10]. To address these concerns, standards must extend beyond technical assay parameters to include documentation requirements, auditability of algorithmic decisions, and mechanisms for ensuring reproducibility across laboratories [4,9].

5. The Need for Dedicated Standards

Given the accelerating integration of AI into molecular diagnostics, standard setting bodies at national and international levels, including ISO and organizations involved in food and health regulation, should prioritize the development of dedicated guidelines and standards [3,4,8]. Such frameworks should encompass not only analytical performance requirements but also data governance, algorithm lifecycle management, and quality assurance principles specific to AI driven systems [3,10]. Establishing harmonized standards in this area would facilitate inter laboratory consistency, support regulatory acceptance, and promote responsible innovation [4,8].

Conclusion

Standardization of artificial intelligence applications in molecular diagnostics is not merely a technical requirement but a fundamental prerequisite for ensuring result reliability, regulatory confidence, and public trust [1–5]. Addressing current gaps in standards will be essential for the safe, transparent, and sustainable integration of AI based technologies into molecular testing in food, agriculture, and public health sectors [3,4,8].

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Consent for publication

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