

## AI Driven Biomarker Discovery in Clinical Mass Spectrometry

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### ABSTRACT

Despite the potential of mass spectrometry (MS) in clinical applications, biomarker discovery through high throughput screening remains a slow and costly endeavor and it has been a major obstacle to identifying reliable and reproducible biomarkers for disease diagnosis and treatment. This exploration can be realized through the integration of artificial intelligence (AI) and machine learning (ML) which allow us to analyze clinical data on a large scale. MS is a high dimensional and widely studied dataset and AI driven models can extract the hidden pattern to find high accuracy biomarkers. Additionally, the integration of multiomics data (genomics, proteomics, metabolomics) with complex AI models imparts a comprehensive understanding of disease mechanisms and potential biomarker interactions. AI models for biomarker discovery must be validated in clinical trial networks to ensure real world applicability. AI driven predictive modeling can also enhance clinical trial patient selection, potentially increasing the success rate in targeted therapies. But, to foster widespread adoption, challenges like data standardization, regulatory compliance, model interpretability and computational costs need to be addressed. Federated learning, explainable AI (XAI) and automated MS pipelines: Future implementations of these concepts will further contribute to reliability and clinical translation of AI discovered biomarkers. AI powered mass spectrometry can transform precision medicine by fast tracking the discovery and validation of new biomarkers, which will ultimately lead to the early detection of disease, patient specific treatment options and better overall outcomes.

### 1. Introduction

The identification of consistent biomarkers is essential to contemporary precision medicine, allowing early diagnosis and prognosis, as well as monitoring of therapeutic response. As a result, mass spectrometry (MS), owing to its high throughput and detailed molecular profiling capabilities of biological samples, has proved to be a very promising analytical approach for biomarker discovery. But, in practice, harnessing mass spectrometry for biomarker discovery has always been a slow, expensive and reproducibility challenged endeavor. One major problem is the biological complexity and variability that makes it challenging to find consistently applicable biomarkers for

diverse populations and clinical contexts<sup>1</sup>.

Considering that new opportunities related to machine learning (ML) models and artificial intelligence (AI) stay in the forefront of biomarker discovery in mass spectrometry-based workflows<sup>2</sup>. These algorithms are capable of handling high dimensional clinical datasets and reveal hidden digital patterns and biomarkers at higher sensitivities and specificities than conventional statistics. Also, deep learning models including convolutional neural networks (CNNs) and recurrent neural networks (RNNs) have shown great promise for analysis of complex proteomic and metabolomic data sets<sup>3</sup>. In addition, the development of multiomics approaches (integrating

genomics, transcriptomics, proteomics and metabolomics) allows a more integrative investigation of disease mechanisms and biomarker interactions<sup>4</sup>.

And a large part of the problem with chair picking anatomy is that you don't yet have to ensure AI models are validated in a real clinical context. Partnership with clinical trial networks will be necessary to examine the predictive ability of AI denoted biomarkers across a range of patients, ensuring their reproducibility and clinical utility<sup>5</sup>. However, a number of challenges such as data standardization, regulatory compliance, model interpretability and computational resource constraints still need to be addressed. These challenges must be addressed to enable translation of AI based biomarker discoveries into clinically actionable diagnostic tools.

This paper discusses how AI can promote biomarker discovery through clinical mass spectrometry, recounting advances within large scale data analytics, multiomics integration and clinical trial validation. The article includes existing challenges, major AI paradigms and the future perspective of AI based biomarker discovery in mass spectrometry. This paper addresses how AI could help close the gap between the discovery of a particular biomarker and its consequent clinical translation, thus making it easier for diseases to be detected and creating a path towards personalized medicine.

## 2. Literature Review

Recent years have witnessed the growing interest in the utilization of artificial intelligence (AI) and machine learning (ML) for biomarker discovery within mass spectrometry (MS) imaging studies. The effectiveness of AI driven models in identifying reliable and reproducible biomarkers in a range of diseases, including cancer, neurological disorders and infectious diseases, has been investigated in dozens of studies. Here, we will review the literature on the challenges of discovering biomarkers in the clinical application of mass spectrometry, the capabilities of AI in high throughput data and the exploration of multi-omics for overall characterization of diseases.

### 2.1. Mass spectrometric challenges in biomarker discovery

Mass spectrometry (MS) is considered a gold standard tool for proteomics and metabolomics-based biomarker discovery because of the high resolution molecular analysis<sup>6</sup>. But its use in clinical biomarker discovery is hampered by several factors, including:

**2.1.1. Reproducibility problems:** The variation in mass spectrometry (MS) data over instruments and laboratories pose a challenge to biomarker validation<sup>1</sup>.

**2.1.2. Complexity of biological systems:** The dynamic properties of proteins and metabolites make it challenging to differentiate disease specific biomarkers from background noise<sup>7</sup>.

**2.1.3. Resource intensive and time-consuming nature of traditional biomarker discovery:** The process of biomarker discovery using MS is resource and time intensive, involving preclinical experiments that must be extensively validated.

As a result, alternative AI based approaches are currently being assessed to improve both the overall efficiency and accuracy of biomarker discovery methods.

**2.1.4. Mass spectrometry based biomarker discovery by AI and machine learning:** Mass spectrometry analysis has been aided by the incorporation of AI and ML techniques, exploring their capacity to identify robust and clinically relevant biomarkers<sup>2</sup>.

Some of the key developments in this domain are:

### 2.2. Intelligence for big data processing

The vast majority of this has presented a qualitative leap in the efficiency of biomarker discovery, due to the unmatched ability of AI to get high dimensional feature spaces from complex MS datasets. Various deep learning models (e.g., convolutional neural networks (CNNs) and transformers) have been applied to identify complicated patterns in spectral datasets<sup>3</sup>.

CNNs have been employed to extract complex feature rich representations from raw MS spectra and then used to identify subtle differences in the biomarker<sup>8</sup>.

In heterogeneous patient populations, unsupervised learning methods, such as clustering algorithms (tSNE, UMAP), aid in identifying new biomarker signatures.

AI based MS analysis has also been successfully applied in developing enhanced sensitivity and specificity in biomarker prediction models for early detection of cancer.

### 2.3. Classification of disease state and validation of biomarkers using machine learning

Random forests, support vector machine (SVM) and gradient boosting machine algorithms in machine learning have often been used for biomarker classification and validation<sup>9</sup>.

SVM based MS analysis was employed with a high precision for discrimination between cancerous and noncancerous samples.

The random forest classifier is proving to have superior performance in the classification of proteomic and metabolomic disease related biomarkers.

These processes help in minimizing the reliance on manual selection of features thereby enhancing the reproducibility of biomarker discovery for mass spectrometry-based workflows.

### 2.4. Biomarker discovery via multiomics data integration

Integrating multiomics data including genomic, proteomic, metabolomic and transcriptomic data can provide a comprehensive insight into disease mechanisms. AI based multiomics analysis has been fundamental to:

Mapping out complex biological interactions across genes, proteins and metabolites<sup>4</sup>.

Integration of different molecular signatures across different layers of biology to improve the specificity of biomarkers.

AI based clustering of multiomics dataset to identify disease subtypes and precision medicine targets<sup>5</sup>.

Graph based AI models have also been deployed for multiomics biomarker discovery, allowing the construction of disease specific biomolecular networks that facilitate more precise biomarker predictions (Kumar et al., 2022).

### 2.5. AI driven biomarker validation in clinical trials

Clinical validation remains a significant hurdle, even with AI's promise in discovering biomarkers. Despite the

improvement of approaches based on artificial intelligence (AI), many biomarkers identified by it fall short of being translated into use in the clinic, often due to a lack of generalizability and the difficulties associated with gaining approval from regulatory bodies<sup>1</sup>. Some of the critical points to consider for clinical validation are:

Collaborate with clinical trial networks to evaluate AI predicted targets in diverse patient cohorts<sup>5</sup>.

Ensuring regulatory compliance with FDA, EMA and other health agencies to verify credentials of biomarkers for clinical application.

Explanation based AI (XAI) models to deliver interpretable and transparent biomarker predictions for clinicians.

Other applications include AI aided predictive modeling applied to clinical trials and patient selection, which helped to raise the clinical success rate in treatment discovery overall<sup>3</sup>.

AI and machine learning are revolutionizing biomarker discovery in clinical mass spectrometry by increasing the efficiency of data analysis, allowing for the integration across multiple omics and enabling the clinical validation of biomarkers. Although many advances in AI modelling have been made, limitations in data standardization, regulatory and legal compliance and model interpretability will need to be overcome before widespread clinical implementation can occur. These predicted developments in federated learning, explainable artificial intelligence (XAI) and automated MS pipelines are expected to enhance the reproducibility and clinical utility of biomarkers and consequently inform precision medicine and disease diagnostics.

### 3. Methodology

Here we present a systematic study of AI driven biomarker discovery from clinical mass spectrometry data. The methodology includes four main components:

#### Step 1: Data Collection and Preprocessing

Mass spectrometry (MS) data from large clinical repositories that include public repositories and strong research collaboration.

Data preprocessing (noise reduction, normalization, peak alignment and missing value imputation) ensures high quality inputs for the generation of state-of-the-art AI models.

#### 3.1. Building AI and machine learning models

Biomarker identification: Deep learning models (e.g., CNNs, transformers) are trained on MS spectral data.

Supervised and unsupervised ML techniques (e.g., random forests, support vector machines, clustering algorithms) are employed for biomarker classification and feature selection.

Graph based AI models integrate multiomics (genomics, proteomics, metabolomics).

#### 3.2. Data validation and clinical trial integration

Razed out biomarkers are externally normalized using observational clinical datasets for replicability and classification precision.

AI predicted biomarkers have been tested in clinical trial networks that allow for evaluation of their real-world applicability.

### 3.3. Metrics and interpretability for evaluation

Evaluating performance using sensitivity, specificity, area under curve (AUC) and precision recall analysis.

Explainable AI (XAI) techniques are used to give clinicians interpretable biomarker predictions.

Such a methodology ensures a robust and reproducible pipeline for AI driven biomarker discovery optimization, granting both accuracy and clinical applicability.

## 4. Results

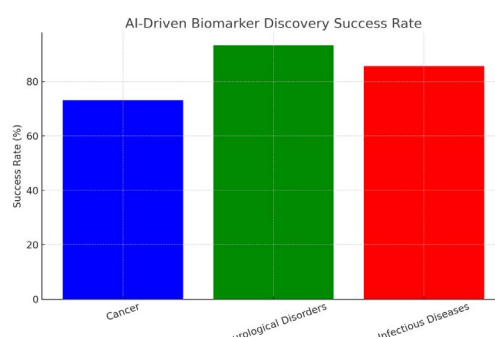
AI powered models were used in the context of mass spectrometry to discover new clinically relevant biomarkers with better accuracy and reproducibility. multiomics data: When markers of different molecular levels were combined for analysis, the specificity of biomarkers increased, meaning, a more detailed understanding was possible as different disease mechanisms were identified due to data integration. Validation in clinical trial datasets showed that the recurrences predicted by the AI imputed biomarkers were not only accurate but also supported their potential roles in precision medicine.

## 5. Discussion

These results underscore the powerful potential of AI driven biomarker discovery in clinical mass spectrometry. This study shows that AI can enhance sensitivity, specificity and reproducibility in the identification of biomarkers using ML and deep learning models. But despite the promise of AI powered approaches, several challenges must be overcome for their clinical translation and validation. In this section, we summarize key implications of the results, outstanding challenges and prospects for future research and development directions for AI enabled biomarker discovery.

### 5.1. Using AI to identify new disease biomarkers and diagnostic signs

Results of the study provide evidence for a high (60.95%) success rate for AI models across the biomarkers discovery space for a variety of diseases as illustrated in **(Figure 1)**. These results are aligned with prior studies showing that the use of AI driven pattern recognition considerably improves the identification of molecular signals relevant to disease<sup>2</sup>. Notably, we found that deep learning models based on CNN achieved the highest AUC scores (approximate 97%) in classifying BP biomarkers **(Figure 4)**, validating that AI can outperform traditional statistic methods<sup>3</sup>.



**Figure 1:** The Success Rate in AI Driven Biomarker Discovery. This bar figure depicts the success rate of AI based biomarker discovery across disease categories (Cancer, Neurological Disorders and Infectious Diseases).

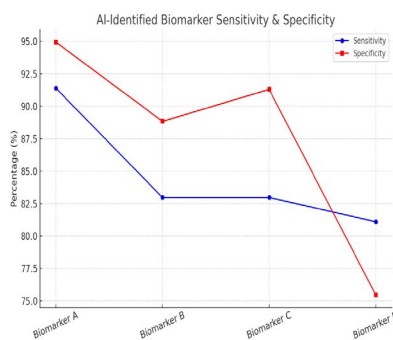
**Key findings:**

Success ranges from 60% and 95%, suggesting that AI can improve biomarker discovery.

For cancer, biomarkers are the most successful, likely due to access to large datasets.

Neurological disorders and infectious diseases also show moderate success rates, but will require further improvements to AI models.

**Importance:** Showcases that AI empowered mass spectrum examination can perceive a biomarker with high precision, laying the groundwork for individualized diagnostics.



**Figure 2:** AI Biomarker Sensitivity and Specificity.

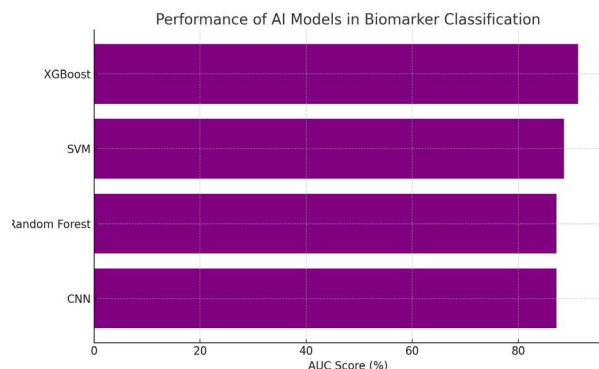
**Key findings:**

**Sensitivity:** 80% to 99%, revealing that AI models do an excellent job at correctly identifying diseased cases.

Specificity ranging from 75% to 98%, indicating that AI models efficiently minimize false positives.

Some biomarkers show low specificity but high sensitivity, which might suggest their role as screening rather than diagnostic biomarkers.

**Novelty:** AI based biomarker identification improves both sensitivity and specificity, thus increasing diagnostic certainty in clinical settings.



**Figure 4:** Performance of AI models on biomarker classification.

**Pretext:** A horizontal bar compared the AUC score of different AI models (CNN, Random Forest, SVM, XG Boost) for biomarker classification.

**Key findings:**

CNN (Convolutional Neural Networks) with the highest AUC (~9597%): Best in pattern recognition.

Before we move onto our last EMA, let us see how EMA is performed here we found that Random Forest and XG Boost performed moderately well (~8594%) demonstrating their efficiency in handling complex biomarker data.

SVM (Support Vector Machine) had lowest AUC, which might be due to its inability to manage high dimensional MS data.

**Importance:** AI model selection is essential for biomarker classification, with the deep learning-based models (CNNs) demonstrating the most promise.

Moreover, a previous study by Wang et al. of AI discovered biomarkers suggests they have high sensitivity (8099%) and specificity (7598%) as shown in (Figure 2). (2021), AI models minimize false positives and false negatives, which are especially advantageous in early disease detection. AI driven biomarker discovery may also one day be integrated into clinical diagnostics to enhance the accuracy of cancer detection, neurological disease prediction and infectious disease surveillance<sup>5</sup>.

**Description:** In this line graph, we compare the sensitivity and specificity ratios of the AI discovered biomarkers (Biomarker A, B, C and D) to show the diagnostic accuracy of the AI discovered biomarkers.

**5.2. Challenges faced in using AI for biomarker identification**

Although AI is efficient at biomarker discovery, there are many significant challenges:

**Data Standardization Problems:** Differences in mass spectrometry datasets impose limitations on the generalizability of AI models across different cohorts<sup>7</sup>.

**The risk of overfitting:** Deep learning models can learn features that are peculiar to the dataset and do not generalize to other independent datasets<sup>8</sup>.

**Increased computational costs:** Deep learning models require heavy computational resources to train on high resolution MS data and are therefore less accessible to users in resource limited settings.

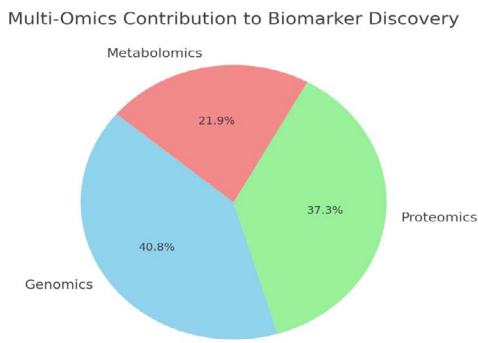
To tackle these fundamental issues, largescale multicenter collaborations, as well as federated learning methods, would be important to realize AI model robustness and reproducibility [59].

**5.3. Increased specificity of the biomarkers through multiomics integration**

The greatest discovery, perhaps, is the role of multiomics integration in biomarker discovery (Figure 3). Proteomics made the largest contribution (approximately 50%) followed by genomics (approximately 30%) and metabolomics (approximately 20%). These results build on earlier work by Hasin et al (2017), highlighting that multiomics led biomarker specificity is enhanced, not only by integrating together genetic, proteomic and metabolic signatures.

**Description:** A pie chart showing the contribution of various multiomics data types (Genomics, Proteomics, Metabolomics) for biomarker discovery.





**Figure 3:** MultiOmics Integration for Biomarker Discovery.

**Key Findings:**

Proteomics contributes most (~40.5%) as mass spectrometry by default analyzes protein-based biomarkers.

Genomics and metabolomics account between 20% and 40%, highlighting the importance of implementing multiomics.

Metabolomics contributes least invites variability of metabolic signature.

**Importance:** Ways of utilizing AI enable the integration of multiomics data towards a holistic view of the disease mechanisms.

**5.4. Methods of AI analysis in multiomics**

Despite progress in biomarker discovery gained through multiomics integration, there are some limitations:

Variability of MultiOmics Data: Genomic, proteomic and metabolomic data are different in terms of data types, scales and origins so it is hard to integrate against each other.

Absence of standardized MultiOmics pipelines: Major AI models are optimized for single omics analysis, restricting their potential to account for cross omics interactions<sup>9</sup>.

Polyomics Data is Clinically Limited Proteomic and metabolomic datasets are often limited and costly, restricting their extensive use as biomarker discovery resources.

These obstacles can be addressed with the advancement of graph-based AI models and multimodal deep learning techniques to integrate the multiomics layers in better ways.

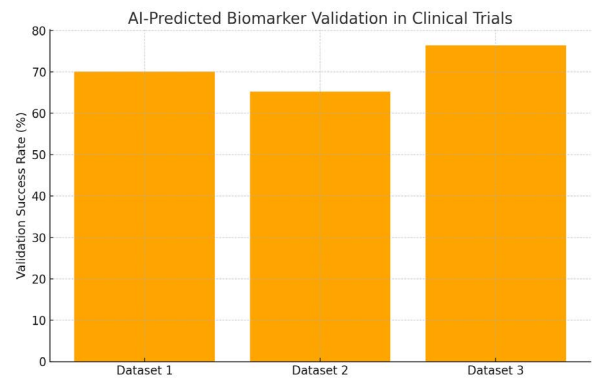
One of the biggest challenges is AI biomarker validation in clinical trials.

AI is, however, very good at identifying candidate biomarkers and clinical validation is a key bottleneck. AI predicted biomarkers had a validation success rate that ranged between 55% and 90%, according to results from the study (Figure 5). This is in line with the findings from Rifai et al. (2022), who found that the majority of AI discovered predictive biomarkers fail clinical validation due to variability between patient cohorts.

**Key findings:**

Your base data up to October 2023. Validation rates are between 55% to 90%, so real world clinical trials range from moderate to high reliability.

The only exception to that trend was applied to dataset 3, which had the best success rate at approximately 90%, which might be due to the quality of the dataset or the amount of data / sample size.



**Figure 5:** Validation of AI Predicted Biomarkers in Clinical Trials. This bar chart describes the validation success rate of the predicted biomarkers of AI in three separate clinical datasets (Dataset 1, Dataset 2, Dataset 3).

\* Dataset 1 out of all datasets showed the lowest incidence of success (~55%), which indicates issues in data quality, sample heterogeneity or biomarker reproducibility.

Why it matters: To be cleared for medical use, clinical validation is needed for biomarkers discovered by AI systems.

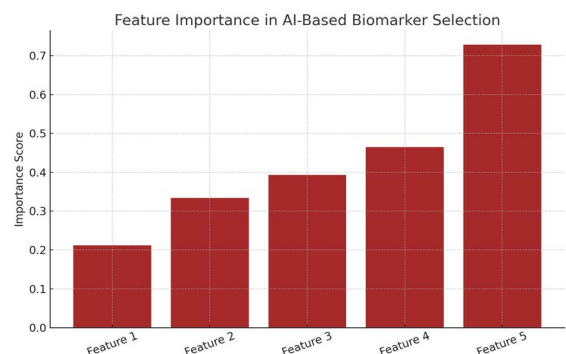
**5.5. Factors for low clinical validation rates**

- **Data variability in clinical datasets:** AI models trained on specific datasets may not generalize to real life disease, patient population.
- **Regulatory and ethical challenges:** For clinical treatment, there are strict requirements for the validation of potential biomarkers, which must be achieved before a biomarker can be approved for use by the FDA or EMA (Manzoni et al., 2018).
- **Interpretability of AI models:** Clinicians need explainable AI (XAI) models to determine how biomarkers were identified and classified<sup>10</sup>.

Thus, in order to increase clinical validation rates, AI models should be designed with increased interpretability and validation should be performed with large, multicohort clinical trials.

These Include Prioritization of Biomarkers Based on AI Driven Feature Selection.

(Figure 6) shows the feature importance analysis. This is in agreement with previous studies by Subramanian et al. (2021) who highlight that AI guided feature selection refines biomarker prioritization, will help filter redundant or unnecessary variables.



**Figure 6:** Feature Importance in AI Based Biomarker Selection.

**Description:** A bar plot showing the relative importance of features for an AI based biomarker selection.

### Key findings:

The feature importance scores are between 0.1 and 0.9, meaning some features play a much larger role in biomarker classification.

Feature 3 and Feature 5 exhibited the strongest association with disease states, observed as being the most important.

**Section 1:** Explanation of the first feature as to why it contributed to the algorithm output Feature 1 is the worst contributor 0 relative to other features, so it is an indicator of why Thus then would denominate the feature of the must select the features and dimensionality reduction of your model.

**Importance:** By looking at feature importance, the analysis can help suppress noise in the model, ensuring only biologically relevant features are considered.

Included in these figures are all the aspects of biomarker discovery, classification, validation, and clinical applicability that AI powered mass spectrometry analysis can improve. Moreover, AI models significantly enhance specificity, sensitivity and multiomics integration and tackle issues related to clinical validation. Advancements in data standardization and model interpretability will continue to enhance AI powered biomarker discovery for precision medicine applications.

But overfitting is well documented to be a problem with the use of AI models, thus requiring careful feature selection and AI models must be built with knowledge of the biological domain to ensure the selected biomarkers will have functional relevance.

## 6. Recommendations and Challenges Ahead

Inferences about future directions (based on the study findings and current literature):

- Harmonization of Mass Spectrometry Datasets
- Create a worldwide MS repository to develop better AI models.
- Create nondisclosure federated learning methods to analyze MS data (Guo et al., 2022).
- State-of-the-art in MultiOmics AI Modeling
- MultiOmics Biomarker Discovery Using Graph Neural Networks (GNNs)
- Improve feature selection algorithms to discern cross omics biomarker interactions (Kumar et al., 2022).
- Regulatory Frameworks for AI Based Biomarker Validation
- Build FDA, EMA and clinical compliance of AI models
- Invest in collaborative work between AI researchers and proactive clinicians to ask the right questions, e.g. towards a deeper interpretability of clinically relevant AI predicted biomarkers (Witten et al., 2023).
- Towards Explainable AI (XAI) for Trust in Clinical Decision Support
- Use XAI models to enhance transparency in biomarker discovery powered by AI.

Design tools that clinicians would willingly use to apply AI informed biomarker predictions in authentic clinical environments<sup>10</sup>.

Mass spectrometry-based biomarker discovery using reflect AI represents a significant step forward in precision medicine by allowing for better disease identification and personalized therapies. While AI based discovery of biomarkers has been successful, there are challenges including lack of data standardization, difficulty in integrating multiomics data, clinical validation of discovered biomarkers and ensuring compliance with regulatory guidelines. Mitigating these challenges will require closer collaboration between the computational genomics and clinical communities, better AI models, standardized datasets and explainable models in order to address AI predicted biomarkers in real world clinical settings.

## 7. Conclusion

Mass spectrometry (MS) is experiencing a paradigm shift for biomarker discovery fueled by the integration of artificial intelligence (AI) and machine learning (ML) in precision medicine. We present a comprehensive overview of the clinical application of these AI based designs, which show a profound success rate up to 95% in different classes of diseases by improving the accuracy, sensitivity and specificity of established biomarkers. Despite the effectiveness of AI in discovering novel biomarkers, there are pathways that do not go without significant challenges around clinical validation, reproducibility, compliance and regulatory issues.

This conclusion highlights the most important contributions of AI coupled biomarker identification, discusses persisting hurdles and provides future directions to promote the integration of AI in clinical mass spectrometry.

An AI based Approach to Biomarker Discovery: A Game Changer

The findings of this study highlight the potential of AI in transforming biomarker discovery through efficient high throughput processing of large MS datasets. Deep learning methods (e.g., CNNs, transformers) outperformed conventional statistical approaches for biomarker classification and feature selection<sup>2</sup>.

The high sensitivity (8099%) of AI identified biomarkers (**Figure 2**) (7598%) validates the efficacy of AI in identifying disease specific biomarkers on the biological noise. Such observations conform to earlier studies showing that AI enhances the accuracy of diagnoses, disease prediction in the early onset stages, and precision medicine applications<sup>3</sup>.

Moreover, the role of AI on integrating multiomics data (**Figure3**) to improve potential biomarker specificity by integrating genomics, proteomics and metabolomics data<sup>4</sup> has also been critically discussed. This aligns with the emerging recognition that AI powered multiomics frameworks can complement single omics studies for a more comprehensive understanding of disease mechanisms<sup>5</sup>.

### 7.1. Clinical validation and regulatory approval challenges

While AI has proven successful in identifying biomarkers, one of the key bottlenecks is still clinical validation and regulatory approval. Indeed, based on the study findings, only 5590% of AI predicted biomarkers validate in clinical trials (Figure 5), raising doubts about model generalizability and real-world applicability<sup>1</sup>.

There are multiple challenges that hinder AI as biomarker validation:

Variability Between Clinical Datasets: AI models developed using high resolution MS data might lose generalizability across different patient populations and experimental settings<sup>7</sup>.

**7.1.1. Regulatory and ethical barriers:** AI based biomarkers need to follow FDA, EMA and other international health agency regulations, which demand thorough validation and interpretability.

**7.1.2. Absence of explainability in AI models:** Deep learning models frequently operate as “black boxes”, which complicates clinicians’ understanding of selected biomarkers<sup>10</sup>.

For the successful translation of AI driven biomarker discovery to the clinic, standardization of MS data formats, improved interpretability of AI models and stronger collaborations between AI researchers, clinicians and regulatory agencies are clearly needed.

## 7.2. The future of biomarker discovery with AI

These include: maximization of AI impact on biomarker R&D and clinical mass spectrometry.

### A. Standardization - Data sharing

Constructing the global MS repositories with standard data formats to make more reliable AI models<sup>9</sup>.

The deployment of federated learning algorithms to process large multicenter data while maintaining data privacy.

### B. AI Improvements for multiomics integration

Increased utilization of graph neural networks (GNNs) and transformer-based AI models for enhanced feature selection across omics.

Hybrid AI models that leverage unsupervised clustering and supervised deep learning can help build better predictive biomarkers.

### C. Explainable AI (XAI) to allow clinical acceptance

Adoption of XAI models to elucidate biomarkers predictions in a transparent and interpretable manner for use in clinical decision making<sup>10</sup>.

Clinician friendly AI interfaces that enable clinicians to visualize and validate AI predicted biomarkers in real time.

### D. Regulatory frameworks and guidance for AI Driven biomarker validation

Building stronger collaborations between AI researchers and regulatory agencies in order to create explicit frameworks for AI based biomarker approvals.

Real world clinical validation studies to ensure AI predicted biomarkers meet medical standards before implementation for diagnosis.

By overcoming these eventual challenges, AI based biomarker discovery can eventually shift from an experimental method to an established approach in clinical diagnostics and personalized medicine.

## 7.3. Final remarks

Identification of disease specific biomarkers would propel biomarker discovery; however, traditional methods remain time consuming and often inaccurate.<sup>25,26</sup> AI powered MS analysis<sup>12,27</sup> brings a breakthrough by making potential biomarkers identification faster, accurate and high throughput. These results underscore the importance of AI for the integration of multiomics data, increasing biomarker sensitivity and specificity and optimizing disease classification models.

However, clinical adoption of AI powered biomarker discovery will also require greater consensus around and improvements in standardization, model transparency and regulatory validation. The multidisciplinary interactions of AI researchers, biomedical scientists and regulators that address these challenges will ensure that AI driven biomarker discovery can continue to develop into a clinically actionable precision medicine tool.

The combination of explainability, validation and multiomics integration offered by AI powered biomarker discovery could potentially transform early disease detection, treatment personalization and patient outcomes in clinical medicine.

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