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Review Article

Biomarkers in health diagnostics: Current perspectives and future directions

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Abstract

Biomarkers have become a cornerstone of modern medicine. They provide measurable signals that reflect normal physiology, ongoing disease processes, or how a patient is responding to treatment. Today, they are applied widely in oncology, cardiovascular disease, infectious illnesses, and neurological disorders. In these areas, biomarkers assist with earlier detection, guide the choice of therapy, and allow clinicians to monitor outcomes over time. Depending on their nature, biomarkers may be molecular (such as DNA mutations or protein levels), imaging-based (such as radiological patterns), or physiological (such as heart rate or glucose levels). They are also classified by purpose, including diagnostic, prognostic, predictive, and monitoring roles.

Although the field has progressed rapidly, moving biomarkers into routine practice is still not straightforward. Many promising candidates fail during large-scale validation because of variability between populations, lack of assay standardization, or the high costs of advanced tests. These limitations are most evident in low- and middle-income countries, where health infrastructure is often limited. Tackling these issues will require consistent validation, clearer regulatory pathways, and the development of affordable, point-of-care technologies.

Looking ahead, new approaches such as multi-omics platforms and advanced computational methods, including artificial intelligence and machine learning, are expected to identify biomarkers with greater accuracy. Non-invasive and wearable biosensors will make monitoring more accessible, even in rural or resource-limited areas. Overall, biomarkers are likely to change the way diseases are diagnosed and managed, but progress will depend on ensuring that these tools are reliable, affordable, and relevant across different populations.

Keywords: Biomarkers, Health Diagnostics, Multi-Omics, Artificial Intelligence, Wearable Biosensors, Personalized Medicine.

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1. Introduction

Biomarkers are measurable biological characteristics that provide information about normal physiology, disease progression, or therapeutic response.[1] Their importance has increased alongside advances in genomics, proteomics, metabolomics, and computational biology, which have accelerated discovery and enabled broader clinical applications.[2,8] Unlike conventional diagnostic methods, biomarkers allow earlier detection of disease, prediction of outcomes, and more personalized treatment planning, making them central to modern precision medicine.

The growing burden of cancer, cardiovascular disease, infectious illnesses, and neurological disorders has further highlighted the need for reliable biomarkers. In oncology, circulating tumor DNA, tumor-associated proteins, and other molecular markers are increasingly applied for early diagnosis, therapy selection, and monitoring.[2,3,5] Infectious diseases also benefit from biomarker research; for example, blood-based biomarker panels are being explored for tuberculosis, which remains a major public health issue in India.[4,6] Similarly, cardiovascular biomarkers such as high-sensitivity troponins, along with digital health tools like continuous glucose monitors, have transformed patient monitoring and risk prediction.[7,10,12] Recent advances in wearable devices and portable biosensors extend these benefits to community and rural settings, offering scalable solutions for long-term health tracking.[8,11]

Despite such progress, translation into routine healthcare is still limited by challenges including high cost, variability across populations, and lack of standardized protocols. This review summarizes the classification and clinical applications of biomarkers, highlights the key challenges restricting their wider use, and discusses future directions involving multi-omics, artificial intelligence,

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and wearable biosensors that have the potential to reshape biomarker-based diagnostics.[9]

2. Types of Biomarkers

Biomarkers can be classified in multiple ways depending on their biological origin and clinical role. Broadly, they may be grouped into molecular, imaging, and physiological/clinical biomarkers, and further subdivided based on their clinical utility as diagnostic, prognostic, predictive, or monitoring tools.

2.1. Molecular biomarkers

Molecular biomarkers include DNA, RNA, proteins, metabolites, and lipids that reflect underlying pathophysiological processes. For example, BRCA1/2 mutations in breast cancer and KRAS mutations in colorectal cancer are widely used in oncology.[5] In the Indian context, HPV DNA testing has been introduced for cervical cancer screening, providing a biomarker-based alternative to cytology and visual inspection, which often face infrastructural challenges.[6,10] Similarly, serum glycated hemoglobin (HbA1c) remains a cornerstone biomarker for diabetes diagnosis and monitoring, highly relevant in South Asia where diabetes prevalence is among the highest globally.[7,11] Advances in multi-omics platforms now enable combined genomic, proteomic, and metabolomic biomarker discovery, offering opportunities for more precise diagnostics.[8,12]

2.2. Imaging biomarkers

Imaging biomarkers are derived from modalities like MRI, CT, PET, or ultrasound. Globally, FDG-PET uptake is used to assess tumor metabolism and response to therapy. In India, imaging biomarkers have been explored for oral cancers, which are highly prevalent due to tobacco use. Radiomics and AI-enhanced imaging tools are being developed to identify early malignant transformations in oral lesions and head-and-neck cancers.[9,12] Such innovations illustrate the growing role of imaging biomarkers in regions with high cancer burden.

2.3. Physiological / Clinical biomarkers

These include measurable clinical or physiological parameters such as blood pressure, heart rate variability, or glucose levels. In Asia, wearable sensors for continuous glucose monitoring are increasingly adopted, particularly in urban diabetic populations.[10,12] Similarly, heart rate and ECG-based wearables are being piloted for atrial fibrillation detection in community health programs in India, offering scalable biomarker-based screening outside hospitals.[11,13]

2.4. Classification by clinical use

Apart from biological type, biomarkers can also be categorized by their clinical application:

1. Diagnostic biomarkers → e.g., HPV DNA test for cervical cancer,[6,14] sputum protein signatures for tuberculosis.[4]
2. Prognostic biomarkers → e.g., HER2 expression in breast cancer predicting aggressive disease course.
3. Predictive biomarkers → e.g., EGFR mutation in lung cancer, guiding use of tyrosine kinase inhibitors.
4. Monitoring biomarkers → e.g., HbA1c for long-term diabetes management,[7,15] cardiac troponins for myocardial infarction recovery.

This functional classification is especially critical in India and Asian countries,where cost-effectiveness and scalability are essential for real-world adoption of biomarker technologies **(Figure 1)**.

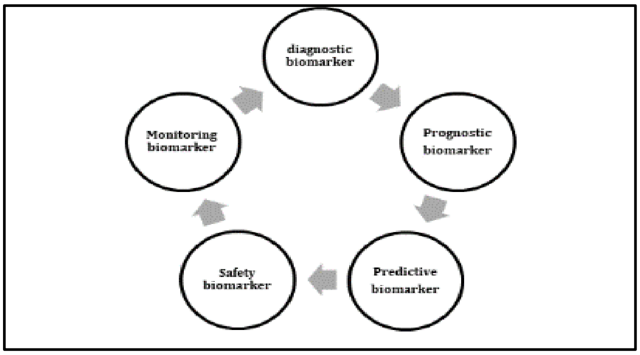

Figure 1: Classification of biomarkers.

3. Applications of Biomarkers in Diagnostics

Biomarkers have a wide range of applications in clinical practice, ranging from oncology and cardiovascular disease (CVD) to infectious and neurological disorders. Their role extends across diagnosis, prognosis, prediction of therapeutic response, and monitoring of disease progress

3.1. Cancer diagnostic

Cancer remains the leading area for biomarker research and clinical application. Circulating tumor DNA (ctDNA), circulating tumor cells, and protein biomarkers are being widely applied in breast, lung, and colorectal cancers.[2,16] In India, HPV DNA testing has been successfully introduced as a screening tool for cervical cancer, reducing mortality in large-scale trials.[6,16] Additionally, HER2 amplification in breast cancer and EGFR mutations in lung cancer serve as predictive biomarkers guiding targeted therapies.[5] The integration of AI with biomarker data and radiomics is further enhancing diagnostic precision for cancers, including oral cancer, which has a particularly high prevalence in South Asia due to tobacco use.[9,14]

3.2. Cardiovascular diseases (CVD)

In cardiovascular medicine, biomarkers are essential for both acute and chronic disease management. Cardiac troponins are the gold standard biomarkers for myocardial infarction, while natriuretic peptides (BNP, NT-proBNP) are used for diagnosing and monitoring heart failure.[11,12] In India, where CVD burden is rapidly increasing, biomarker-based screening has significant potential for early detection. Emerging biomarkers such as high-sensitivity C-reactive protein (hs-CRP) are also being investigated for cardiovascular risk stratification. Wearable devices that track ECG and heart rate variability are extending the utility of biomarkers into real-world monitoring, as demonstrated in the mSToPS trial.[11]

3.3. Infectious diseases

Biomarkers are also critical in infectious disease diagnostics. For tuberculosis (TB), blood-based host immune and metabolic signatures are being explored to distinguish

active from latent infection.[4] In India, where TB remains endemic, such biomarkers could transform early diagnosis and treatment initiation. During the COVID-19 pandemic, biomarkers such as IL-6, D-dimer, and ferritin were used for prognosis and severity prediction.[13] These examples highlight the value of biomarkers in improving outcomes for infectious diseases, especially in resource-limited settings.

3.4. Neurological disorders

Neurological diseases, particularly Alzheimer's disease (AD) and Parkinson's disease (PD), are another domain where biomarkers are increasingly applied. In AD, amyloid-beta and tau proteins in cerebrospinal fluid (CSF) and more recently in blood, are recognized diagnostic biomarkers.[14] In PD, biomarkers such as alpha-synuclein and neurofilament light chain (NfL) are being investigated to support early diagnosis. In Asia, where dementia prevalence is rising, biomarker-based approaches could significantly improve early intervention strategies **(**

Table 1)

AI-enhanced neuroimaging biomarkers are also showing promise in detecting structural brain changes associated with these disorders.

4. Challenges

Despite significant progress, the translation of biomarkers from discovery to clinical practice faces multiple challenges. These barriers exist across scientific, technical, regulatory, and socioeconomic domains **(Figure 2)**.

Disease	Biomarkers	Clinical Applications
Oncology	HER2/neu, BRCA1/2, EGFR, ALK	Targeted therapy guidance, diagnosis, monitoring
Infectious Diseases	IL-6, CRP, D-dimer, CD4, Viral load	Early diagnosis, severity prediction
Cardiovascular diseases	Troponins, BNP/NT-pro BNP	MI detection, heart failure prognosis
Metabolic Disorders	HbA1c, FPG, Insulin, C-peptide	Diabetes diagnosis and monitoring
Neurological Disorders	Beta-amyloid, tau, alpha-synuclein	Alzheimer's/Parkinsons diagnosis, tracking
General Drug Response	Blood pressure, INR, Viral load reduction	Drug monitoring, Safety/efficacy

Table 1: Representative biomarkers across major disease areas and their clinical application.

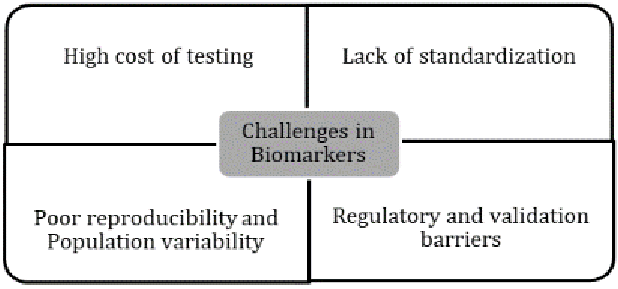

Figure 2: Key challenges in Biomarkers translation

4.1. Scientific and technical challenges

Biomarker discovery often begins with promising results in controlled research environments, but reproducibility across diverse populations remains limited. Many candidates fail during large-scale validation due to genetic diversity, comorbidities, and environmental differences.[1] Moreover, the sensitivity and specificity of biomarkers may decline in real-world clinical settings compared to laboratory conditions.[5]

4.2. Standardization and validation issues

There is a lack of standardized protocols for sample collection, storage, and assay methods, leading to variability in results between laboratories.[15] This makes it difficult to compare findings across studies or establish universally accepted cutoff values. In India, infrastructural gaps such as inconsistent laboratory quality control and limited access to advanced molecular assays exacerbate these challenges.

4.3. Regulatory and ethical barriers

The regulatory pathway for biomarker approval is complex and often varies across countries. Rigorous validation is required before biomarkers can be accepted for clinical use, which is time-consuming and expensive.[16] Ethical concerns, such as privacy and data protection in AI-driven biomarker platforms, also need careful consideration, particularly with genetic biomarkers.

4.4. Cost and accessibility

The development and implementation of biomarker-based diagnostics can be prohibitively expensive, limiting accessibility in resource-constrained settings. For instance, while next-generation sequencing (NGS) and multi-omics platforms offer powerful insights, their high costs restrict widespread use in low-income countries.[8] In India, even routine biomarker tests such as HbA1c or HPV DNA may remain inaccessible in rural populations due to cost and infrastructural barriers.[6,7]

4.5. Integration into healthcare systems

Even when effective biomarkers are available, integrating them into existing healthcare systems remains a challenge. Lack of trained personnel, limited awareness among healthcare providers, and fragmented health infrastructure reduce their adoption. AI-enabled diagnostic biomarkers require robust digital health frameworks, which are still under development in many parts of Asia.[9]

5. Future Perspectives

The future of biomarker research and application is shaped by technological innovation, precision medicine, and the need for affordable, scalable solutions in both developed and developing regions. Several emerging trends are expected to define the next generation of biomarker-driven healthcare

5.1. Multi-Omics integration and systems biology

Combining genomics, transcriptomics, proteomics, and metabolomics is expected to provide a more holistic understanding of disease mechanisms. These multi-omics approaches allow for the identification of biomarker panels rather than single markers, improving accuracy and reliability.[8] In India, efforts are ongoing to establish population-specific omics databases, which will make biomarker discovery more relevant for regional genetic diversity.

5.2. Artificial intelligence (AI) and machine learning

AI-driven approaches are expected to revolutionize biomarker discovery and application. By analyzing large and complex datasets, AI can identify hidden biomarker signatures and enhance predictive modeling for early diagnosis.[3] Radiomics and digital pathology combined with AI will also enable automated, highly sensitive image-based biomarkers, especially for cancers and neurological disorders.[9]

5.3. Non-Invasive and wearable biomarkers

Future diagnostics will increasingly rely on non-invasive biomarkers such as saliva, urine, breath, and sweat, which are easier to collect and more acceptable to patients.[1] Wearable biosensors capable of continuous monitoring of glucose, cardiac rhythm, or stress-related biomarkers will extend diagnostic reach beyond hospitals. [10,11] In India, the integration of low-cost biosensors into community health programs may significantly improve early disease detection in rural areas.

5.4. Personalized and precision medicine

Biomarkers will play a central role in personalized medicine, enabling treatments tailored to individual patients based on genetic, molecular, and lifestyle profiles. Predictive biomarkers will guide therapy selection, ensuring maximum benefit and minimal toxicity.[5] Advances in pharmacogenomics are expected to make personalized treatment strategies feasible in oncology, cardiovascular disease, and infectious diseases.

5.5. Global and regional collaborations

For biomarkers to achieve clinical impact, strong global and regional collaborations are needed. Large-scale biobanking initiatives, standardized protocols, and open-access databases will accelerate discovery and validation. In low- and middle-income countries like India, collaborations with international research networks can bridge infrastructural gaps, making biomarker-based diagnostics more equitable and accessible.[6]

6. Conclusion

Biomarkers have become essential in modern healthcare, supporting early detection, diagnosis, prognosis, and therapeutic monitoring across multiple diseases. Their role is especially evident in cancer, cardiovascular diseases, infectious illnesses, and neurological conditions, where they guide clinical decision-making and improve outcomes. Despite rapid scientific advances, wider clinical adoption remains limited by high costs, variability across populations, and the lack of standardized protocols.

Future development should focus on robust validation, regulatory alignment, and the creation of affordable, point-of-care tools suitable for diverse healthcare settings. Emerging technologies, including multi-omics, artificial intelligence, and wearable biosensors, offer promising solutions to overcome existing barriers. With continued research and equitable implementation, biomarkers are poised to play a central role in advancing personalized medicine and improving population health worldwide

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