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REVIEW PAPER

CANCER BIOMARKERS AND HEALTH SYSTEMS: THE DYNAMICS OF A SCIENTIFICALLY BASED TRANSFORMATION

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Summary

Cancer biomarkers are molecular markers that play a crucial role in the diagnosis, treatment, and monitoring of cancer. These markers enable more accurate patient classification and the development of personalized treatment strategies. The effect of genetic differences on the disease process necessitates the use of biomarkers in clinical decision-making. Advanced analysis technologies and multi-biomarker panels have increased diagnostic accuracy and facilitated the monitoring of treatment responses. Integrating biomarkers into healthcare policies expands opportunities for early diagnosis, enables more effective management of treatment processes, and increases the cost-effectiveness of healthcare services. Advanced technologies, such as artificial intelligence, nanotechnology, and big data analysis, enhance the sensitivity and specificity of biomarkers, thereby strengthening the success of precision medicine applications. These innovations enable the development of personalized treatment approaches and contribute to the sustainability of healthcare systems. In this context, the integration of biomarkers into health policies expands early diagnostic opportunities, improves service quality, and contributes to the creation of sustainable health systems. It is critically important for policymakers to consider scientific accuracy, patient safety, and ethical principles when evaluating these technological advances in order to effectively manage future health issues.

Keywords: personalized medicine, molecular diagnostic, healthcare services, biomarkers, cancer

Introduction

Cancer is a complex disease that begins and progresses due to numerous and variable genetic mutations. Currently, one in five people worldwide will develop cancer during their lifetime. Consequently, cancer has become a significant public health issue on a global scale [1]. The complex nature of cancer affects not only the development of the disease but also the clinical differences between individuals. In this context, even patients with the same type and stage of cancer may exhibit different prognoses or treatment responses during the course of their disease. In this regard, the discovery and clinical application of biomarkers play an important role in understanding cancer, classifying patients, and improving patient survival rates. In recent years, the use of high-throughput technologies such as next-generation sequencing and mass spectrometry has significantly contributed to the identification of cancer biomarkers at both the proteomic and genomic levels [2,3].

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Aim of the work

This study aimed to comprehensively examine the effects of cancer biomarkers on diagnosis, treatment, and health policies. The contribution of biomarkers to understanding cancer at the molecular level, their role in personalized medicine, and the process of integrating them into health systems are discussed, and the clinical and societal benefits of these technologies are evaluated. In addition, the potential effects of biomarker-based approaches on the sustainability and effectiveness of healthcare services were examined.

Methods

This narrative literature review was conducted using a structured search strategy to ensure comprehensive coverage and methodological clarity. Peer-reviewed publications published between 2008 and 2025 were comprehensively searched in the PubMed, ScienceDirect, Wiley Online Library, Web of Science, and Google Scholar databases. Priority was given to studies published within the last five years (2020-2025). The literature search was conducted using combinations of the following keywords: "cancer biomarkers", "molecular diagnosis", "health systems", "health services", "healthcare systems", "healthcare services", and "personalized medicine". The inclusion criteria encompassed peer-reviewed research and review articles that addressed cancer biomarkers for diagnostic, prognostic, monitoring, or therapeutic purposes, as well as studies examining the implications of these biomarkers in clinical applications and health systems. Editorials, commentaries, conference abstracts, and publications that lacked a scientific peer-review process were excluded.

The article selection process was conducted in two stages. In the first stage, titles and abstracts were screened for relevance, resulting in the preliminary selection of studies. In the second stage, full-text articles were reviewed in detail to assess their eligibility based on the predefined inclusion criteria. Initially, 257 records were identified. After the removal of duplicate entries, titles and abstracts were screened, and 138 full-text articles were selected for a detailed evaluation. Following a comprehensive eligibility assessment, 47 full-text articles were deemed suitable for inclusion in the final analysis. These studies are the core references cited in this review. Data obtained from the selected articles was systematically organized and narratively synthesized to highlight recurring themes, methodological patterns, and biomarker-related insights.

All authors have been actively involved in conducting the literature review, analyzing the data, and interpreting the findings.

Literature review results

Functional categories and applications of cancer biomarkers

Cancer biomarkers are biological molecules, such as nucleotides, proteins, lipids, and metabolites, produced by tumors or tissues in response to cancer. They can be used in clinical settings for diagnosis, screening, prognosis, monitoring treatment efficacy, identifying cancer subtypes, and predicting patient response to specific treatments [4,5]. Based on their biological origin, biomarkers can be classified into tissue, circulating (e.g. circulating tumor DNA), and cellular (e.g. circulating tumor cells) types. Each class of biomarkers provides distinct information on cancer. For example, circulating biomarkers enable non-invasive sampling and offer insights into tumor progression [4,6].

Despite extensive research, only a limited number of biomarkers have entered widespread clinical use because of challenges in validation and the transition from research to clinical application. These biomarkers play diverse roles in cancer biology, including diagnosis, prognosis, and treatment. For instance, Prostate-Specific Antigen (PSA) is widely used for prostate cancer screening, although its diagnostic specificity remains debated. Cancer Antigen 125 (CA-125) is employed in ovarian cancer, particularly for monitoring disease progression but is limited by low sensitivity in early stage detection. Human Epidermal Growth Factor Receptor 2 (HER2) is a critical biomarker for breast and gastric cancers that guides targeted therapy decisions. BRCA1/BRCA2 mutations are associated with hereditary breast and ovarian cancers and have implications for both risk assessment and therapeutic strategies in these cancers. KRAS mutations are common in colorectal cancer and influence the response to anti-EGFR therapies. Other biomarkers, such as Carcinoembryonic Antigen (CEA) and Cancer Antigen 15-3 (CA15-3) are used for monitoring breast cancer. While these biomarkers are integral to clinical practice, their diagnostic sensitivity, specificity, and biological relevance vary, underscoring the need for continued refinement and validation [7,8].

Currently, the determination of cancer biomarkers in clinical practice involves the examination of samples obtained from various body fluids or cellular components using advanced analytical platforms such as polymerase chain reaction (PCR), microarray, next-generation sequencing (NGS), mass spectrometry (MS), and high-performance liquid chromatography (HPLC) [9-14]. Building on these technologies, recent developments in molecular diagnostics and bioinformatics have led to a more nuanced classification of biomarkers based on their clinical roles and technological integration. These biomarker categories (preventive/predictive, diagnostic, monitoring, and therapeutic), along with their core features, recent advances, and associated analytical platforms, are summarized in Table 1.

Table 1. Categories of cancer biomarkers and associated technological advances

Biomarker category	Functional attributes	Recent advances	Technologies and analytical platforms
Preventive / predictive biomarkers	• Identify individual susceptibility and genetic predisposition (e.g. BRCA1/2 variants for breast/ovarian cancer) [15] • Predict therapeutic response via tumor genomic signatures [16] • Enable molecular risk stratification integrating family history and multi-omics profiles [15] • Guide prophylactic or surveillance strategies in high-risk individuals [15]	• Integration of multi-omic data into AI-based risk models [17,18] • Microbiome and epigenome inclusion for personalized risk prediction [17] • Polygenic risk scoring for rare tumors [18] • Improved individualized prevention strategies [16]	• Targeted gene panels and whole-exome/genome sequencing (NGS) • SNP and variant analyses, polygenic risk scoring • DNA methylation and epigenomic profiling • Multi-omic integration with bioinformatics/AI modeling [17,18,27]
Diagnostic biomarkers	• Liquid biopsy for early, non-invasive detection using ctDNA/ctRNA [19,20] • circRNA and miRNA panels for tumor subtyping [21] • Validated molecular assays (e.g. HER2 amplification, SEPT9 methylation) [15] • Imaging biomarkers (FDG-PET/CT, 68Ga-FAPI PET, multiparametric MRI) [22,23]	• Rapid miRNA/exosome-based diagnostic assays [20,24] • Expanded ctDNA panels with improved sensitivity [19] • Electrochemical and optical biosensors [24] • Radiomics-based AI interpretation of medical imaging [23,25,27]	• Digital PCR (ddPCR) and next-generation sequencing (NGS) • Exosome isolation and proteomic characterization • Mass spectrometry-based proteomics • Point-of-care biosensor devices • Radiomics and AI-driven image analysis [18,23-25,27]
Monitoring biomarkers	• Minimal residual disease (MRD) tracking via ctDNA kinetics [19,28] • Dynamic therapy response through exosomal and miRNA changes [16,20] • Quantitative MRI/PET metrics for treatment evaluation [22,23] • Early relapse prediction via serial molecular measurements [19]	• Ultra-sensitive ctDNA assays with expanded variant panels [19] • Real-time point-of-care biosensor monitoring [24] • AI-assisted longitudinal trend and relapse prediction [25,27] • Multi-biomarker integration for enhanced accuracy [19,20]	• Deep NGS and ddPCR/BEAMing approaches • Exosome profiling and serial sampling • Continuous biosensor readouts • Radiomic and machine-learning integration for monitoring [18,24,25,27]

Therapeutic biomarkers	• Approved molecular targets (e.g. HER2 amplification, PD-L1 expression) [15,26] • Emerging targets such as EZH2 and other epigenetic regulators [5] • Resistance mechanism markers and drug sensitivity predictors [16] • Companion diagnostics for biomarker-matched therapies [5,17]	• Biomarker-driven clinical trials and drug matching [5,17] • New generation of epigenetic inhibitors [5] • AI-enhanced translational platforms linking clinic and lab [17,27] • Immunoprofiling-based combination therapy designs [26]	• Clinical-grade NGS panels and mutation detection • Immunohistochemistry and quantitative protein assays • RNA expression and functional genomic signatures • High-throughput drug screening and translational platforms [5,16,18,27]

The growing awareness of the clinical roles of biomarkers has accelerated research aimed at enhancing their sensitivity and specificity. These efforts increasingly position biomarkers not only as diagnostic tools but also as integral components of clinical decision-making across the continuum of cancer care [6,7].

As cancer biomarkers are increasingly implemented in clinical practice, their evaluation requires more comprehensive criteria. This assessment involves factors such as the intended use of the biomarker, its clinical benefits compared to existing alternatives, reliability, cost-effectiveness, societal acceptance, and its potential for early diagnosis. Furthermore, the ability of biomarkers to stratify patients for personalized treatment plays a key role in clinical decision making. However, performance metrics, such as sensitivity and specificity, may be influenced by individual factors, including patient age and disease stage [29].

Biomarkers play a central role in the diagnosis, prognosis, and treatment of diseases in modern biomedical research. Particularly in heterogeneous diseases, such as cancer, inter-individual variability in biomarkers has accelerated the development of personalized therapies. This diversity has highlighted the limitations of single biomarkers in clinical practice and has led to the widespread adoption of multibiomarker panels [5,7,30].

These panels facilitate a more detailed molecular understanding of the disease, enhance the prediction of treatment responses, and enable comprehensive mapping of an individual's biological profile. By integrating genomic, transcriptomic, proteomic, and metabolomic data, health systems can incorporate genetic predispositions, environmental exposure, and lifestyle factors into clinical decision-making. This holistic approach forms the foundation of personalized medicine and supports the development of tailored healthcare solutions [5,30,31].

Building on this foundation, the next section explores how cancer biomarkers influence healthcare delivery, including their role in shaping policies, improving system efficiency, and advancing equitable access to precision care.

Health systems and policy implications of cancer biomarkers

Biomarker-based molecular diagnostics and personalized medicine applications significantly influence health policies, particularly in cancer treatment. Biomarkers facilitate disease identification, prognosis determination, and treatment prediction, enabling early diagnosis and facilitating the development of clinical strategies and shaping clinical practices. These developments have necessitated the updating of health policies to include molecular testing in clinical practice and the development of strategies that enable policymakers to adapt more quickly and effectively to diagnostic innovations [32,33].

The integration of cancer biomarkers into healthcare processes, built upon this transformation, has further deepened their impact on health policies. Particularly in malignancies with a high incidence, such as lung and breast cancer, personalized treatment approaches using proteomic and genomic data are increasingly being developed and are emerging as decisive tools in the clinical decision-making process. This strategic shift in orientation not only increases diagnostic and therapeutic accuracy but also necessitates the creation of new policies aimed at improving access to biomarker-based technologies, reducing associated costs, and aligning legal frameworks with scientific advances [34].

In this context, technologies for the discovery and development of biomarkers are bringing significant changes to the clinical management of various types of cancer, strengthening both personalized treatment approaches and evidence-based practices. For example, advances in proteomics and genomics have enabled the identification of numerous potential biomarkers that support early cancer diagnosis, facilitate accurate prognostic assessment, and enable monitoring of treatment responses. These innovations play an important role in optimizing patient outcomes and reducing the economic burden on health systems [35-38]. Recognizing the transformative potential of these technologies, research and development efforts focused on biomarker innovation have catalyzed policy initiatives aimed at their integration into clinical practice [39]. Such policies contribute to enhancing healthcare quality by promoting harmonized, evidence-based, and scientifically validated diagnostic standards. Moreover, for the effective management of future health challenges, policymakers must evaluate these technological advances through the lens of scientific rigor, patient safety, and ethical responsibility [32,40].

Health strategies aimed at integrating cancer biomarkers into clinical protocols should be based on robust clinical evidence to increase cost-effectiveness, improve service accessibility, and facilitate regulatory approval processes, in line with policy requirements. In this regard, the integration of artificial intelligence-based analytical systems with nanomaterial-based biosensors has the potential to strengthen clinical validation processes by increasing the sensitivity and specificity of biomarker detection. These technological innovations can play a significant role in reshaping the direction and priorities of health policies by accelerating the adoption of personalized and precision medicine approaches [6,41].

On the other hand, the integration of biomarker technologies into healthcare systems has also presented certain challenges. These challenges include increased financial burden, the need for standardized diagnostic and treatment protocols, and the requirement to ensure the clinical validity and utility of the tests. These issues are particularly pronounced in developing countries, such as India. Limited resources, infrastructure deficiencies, and high costs in these countries make it difficult for health policies to adapt to this transformation. The current situation creates significant inequalities in access to molecular diagnostics, requiring policymakers to address this area with inclusive, sustainable, and equitable solutions [34,42].

Despite these challenges, the impact of cancer biomarkers on health management has become increasingly evident, particularly in the context of precision medicine. Biomarkers are central to personalized treatment strategies in oncology and function as measurable indicators of normal or pathological biological processes. For example, in non-small cell lung cancer (NSCLC), recent advances in genomic data and bioinformatics have enabled the use of biomarkers for early diagnosis and personalized treatment planning in NSCLC. Their integration into precision medicine has significantly improved patient management by tailoring treatments to specific disease profiles [43]. Furthermore, these developments have contributed to a strategic shift towards more integrated health models and enabled oncologists to classify neoplastic diseases not only by clinical phenotype but also by molecular characteristics [5]. The multifaceted roles and systemic integration of cancer biomarkers in precision medicine and health system are illustrated in Figure 1.

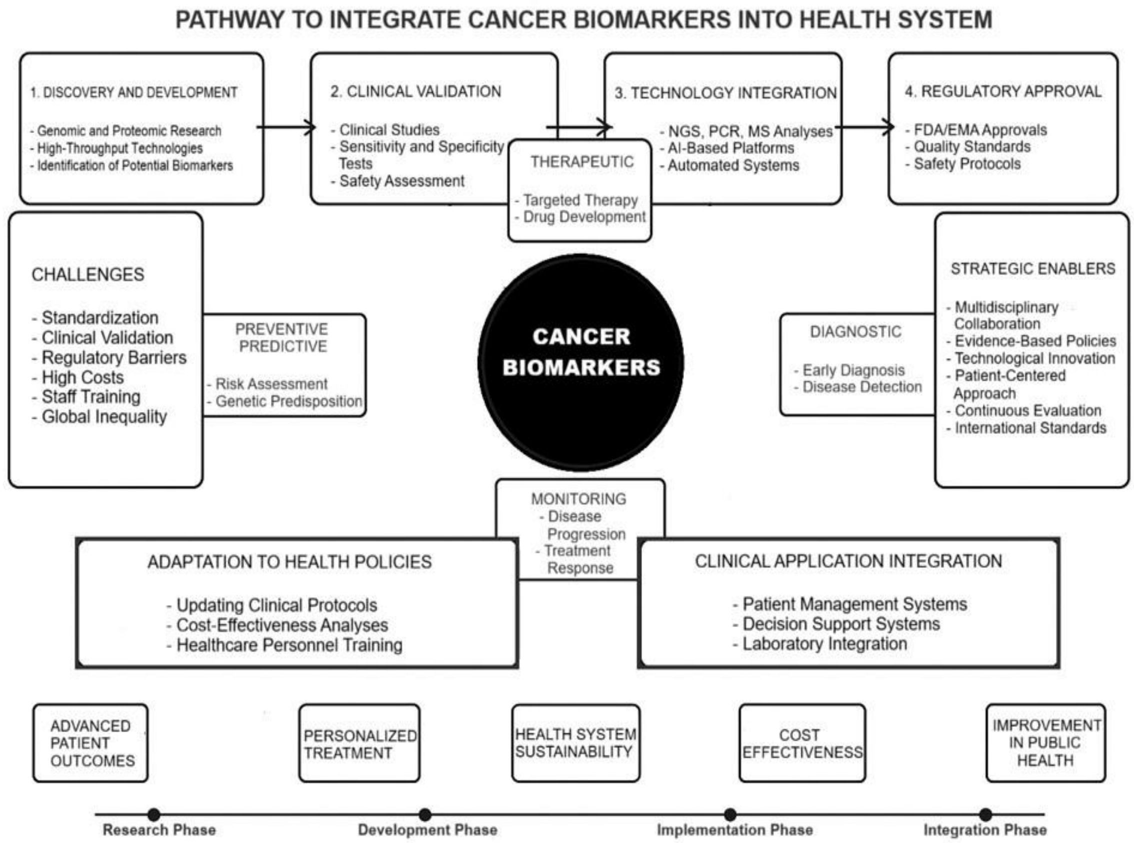

Figure 1. A comprehensive roadmap illustrating the multi-phase integration of cancer biomarkers into health systems

In recent biomedical research, biomarker studies are at the forefront of the field; particularly in complex diseases such as cancer, the emergence of individual differences has increased the importance of multi-biomarker panels beyond single biomarkers [44]. This approach enables more precise subclassification of diseases and the development of personalized treatment strategies. The concept of personalized medicine should be supported by integrated multi-omic systems that encompass not only genetic and molecular data but also an individual's lifestyle, environmental interactions, and dietary habits. This integrated approach, which forms the basis of smart health systems, combined with artificial intelligence and advanced bioinformatics analysis, enables real-time monitoring and analysis of an individual's health data throughout their life cycle [45]. This brings preventive medicine to the forefront, minimizes trial-and-error-based treatment processes, and reduces the economic burden on the healthcare system.

In terms of the sustainability of current health systems, the creation of personalized biological maps and the support of big data analysis using digital technologies have become inevitable in recent years. This transformation will not only improve the quality of life of individuals but also provide new perspectives on drug development processes, thereby enabling more effective management of healthcare expenditures [46,47].

Discussion of the review results

Cancer biomarkers represent a transformative force in modern oncology, fundamentally reshaping healthcare delivery from early diagnosis to treatment monitoring and drug development [1-3]. These molecular markers have evolved beyond simple diagnostic tools to become integral components of personalized medicine, enabling clinicians to understand the biological foundations of cancer while facilitating the development of targeted therapeutic approaches [4-6].

The clinical implementation of biomarkers such as CA-125, PSA, HER2, and BRCA1/BRCA2 mutations has demonstrated the potential and limitations of single-biomarker approaches [7,8]. Although these biomarkers have yielded successful results in clinical applications, variability in diagnostic performance necessitates the development of more reliable and comprehensive biomarker panels. In this context, genomic, proteomic, transcriptomic, and metabolomic data is important [5,30,31]. This evolution toward comprehensive molecular profiling enhances treatment precision by enabling better patient selection for clinical trials, predicting treatment response, detecting drug resistance, and monitoring therapeutic progress [12-14]. The use of advanced analytical methods, such as PCR, NGS, microarray, MS, and HPLC, has facilitated these developments in clinical practice [9-11].

The integration of biomarker technologies into healthcare systems has necessitated significant policy adaptations, particularly in balancing innovation adoption with cost-effectiveness and accessibility [32,33]. The shift toward precision medicine has improved patient management effectiveness while reducing unnecessary drug use and adverse effects, ultimately increasing patient satisfaction and healthcare efficiency [34]. However, implementation challenges persist, particularly regarding global health disparities and the need for standardized diagnostic protocols [34,42]. These challenges are especially pronounced in developing countries, where limited resources, infrastructure deficiencies, and high costs create significant inequalities in access to molecular diagnostics.

The sustainability of modern healthcare systems increasingly depends on the development of integrated multi-omic approaches supported by artificial intelligence and advanced bioinformatics analyses [6,41,45]. The emergence of smart health systems that incorporate real-time health data monitoring throughout an individual's life cycle promises to prioritize preventive medicine while minimizing trial-and-error treatment approaches [45]. This transformation has the potential to reduce the overall economic burden on health systems while improving the individual's quality of life [46,47]. Recent advances in the multi-omics field have enabled the identification of numerous potential biomarkers for the early diagnosis of cancer, determination of disease prognosis, and monitoring of treatment response [37,38].

Conclusions

Cancer biomarkers have multifaceted and profound impacts on health policy, contributing to healthcare systems across a wide spectrum, from early disease detection to the development of next-generation oncology drugs. The effective integration of biomarker discovery and development technologies into clinical practice not only improves the quality and accessibility of cancer care but also supports the sustainability of the health system.

It is critical for policymakers to consider the scientific, economic, and ethical dimensions when evaluating these innovations to effectively manage future health challenges. The rapid and effective integration of advanced cancer biomarkers into health systems will benefit individual patients and enable the delivery of more effective, equitable, and sustainable healthcare services at the societal level. Success in this endeavor requires addressing the current challenges in clinical validation, ensuring equitable global access, and developing inclusive policies that bridge the gap between technological advancement and practical implementation. Therefore, the continued evolution of cancer biomarkers represents not only a technological advancement but also a fundamental shift toward precision medicine that demands coordinated efforts among researchers, clinicians, policymakers, and healthcare systems to realize its full potential for improving global cancer care.

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