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THE ROLE OF EARLY DETECTION AND MOLECULAR BIOMARKERS IN IMPROVING BREAST CANCER SURVIVAL RATES: A REVIEW

Dallo Salva Faekovna
Assistant, Department of Oncology, Oncohematology, and Radiation Oncology,
Tashkent State Medical University, Tashkent, Uzbekistan

Abstract
Breast cancer remains the most prevalent malignancy among women worldwide, accounting for a significant proportion of cancer-related morbidity and mortality. Despite substantial advances in oncology, survival outcomes continue to depend heavily on early detection and molecular profiling of tumors. Early screening methods such as mammography, ultrasound, and MRI have significantly improved diagnostic accuracy and allowed detection at earlier, more treatable stages.
In parallel, molecular biomarkers — including BRCA1/2, HER2, PIK3CA, TP53, and hormone receptor status (ER, PR) — have transformed breast cancer classification and management. These markers provide crucial insights into tumor aggressiveness, therapeutic responsiveness, and patient prognosis. Integrating molecular biomarker assessment with effective early detection strategies allows for precision oncology, leading to personalized treatment and improved survival rates.
This review summarizes current research on early detection approaches and key molecular biomarkers in breast cancer, emphasizing their combined role in enhancing prognosis and tailoring patient-specific therapies.

Keywords: Breast cancer; early detection; molecular biomarkers; BRCA1/2; HER2; hormone receptors; personalized therapy; survival rate.

Introduction

Breast cancer is the most commonly diagnosed malignancy and the leading cause of cancer-related death among women globally. According to the World Health Organization (WHO), more than 2.3 million new cases are registered annually, and approximately 685,000 women die from the disease each year. Despite improvements in surgical techniques, chemotherapy, radiotherapy, and hormonal treatment, the prognosis of breast cancer still depends primarily on the stage at which the disease is detected and the molecular characteristics of the tumor.
Early detection plays a crucial role in reducing mortality and improving quality of life. Regular screening programs using mammography, ultrasound, and magnetic resonance imaging (MRI) enable the identification of tumors at the pre-invasive or early invasive stages, when treatment outcomes are significantly better. Public awareness, accessibility to screening services, and genetic counseling have also contributed to earlier diagnosis in many countries.
At the same time, advances in molecular biology have revolutionized the understanding of breast cancer pathogenesis. Molecular biomarkers such as BRCA1 and BRCA2 gene mutations, HER2

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overexpression, PIK3CA, TP53 alterations, and hormone receptor (ER/PR) status have become essential for classifying tumors, predicting prognosis, and guiding therapeutic decisions. These biomarkers not only reflect the biological behavior of the tumor but also determine sensitivity to targeted or hormone-based therapies.

Integrating early detection methods with molecular diagnostics enables a personalized approach to breast cancer management. Such a strategy allows clinicians to optimize treatment regimens, minimize unnecessary toxicity, and significantly improve overall survival rates. Therefore, understanding the combined role of early detection and molecular biomarkers is of paramount importance for modern oncology and public health.

The Role of Early Detection

Early detection of breast cancer remains one of the most effective strategies to reduce mortality and improve patient outcomes. Numerous clinical studies have confirmed that diagnosis at the initial stages (stages I–II) significantly increases the five-year survival rate, reaching up to 90–95%, compared with less than 30% in advanced cases.

The cornerstone of early detection is **screening**, primarily through **mammography**, which is currently the most widely used and evidence-based method. Mammographic screening allows the identification of microcalcifications and small non-palpable lesions, which may represent early forms of ductal carcinoma in situ (DCIS) or invasive cancer.

In addition to mammography, **ultrasound examination** and **magnetic resonance imaging (MRI)** play essential roles in the diagnostic process, especially for women with dense breast tissue or genetic predisposition. Ultrasound is often used as an adjunct method to clarify suspicious mammographic findings, while MRI provides superior sensitivity in detecting multifocal and multicentric lesions.

Public health programs aimed at increasing awareness and accessibility to screening have demonstrated remarkable results in many countries. Organized screening initiatives, regular check-ups, and education campaigns help women recognize the importance of early diagnosis and seek medical attention promptly.

Furthermore, **self-examination** and **clinical breast examination (CBE)** remain valuable tools, particularly in regions with limited access to advanced imaging technologies. Although these methods are less sensitive than imaging, they are crucial in resource-limited settings for raising awareness and promoting timely detection.

In summary, the integration of various screening modalities and awareness programs contributes to earlier diagnosis, more conservative treatments, and improved survival rates. Early detection not only saves lives but also reduces the physical, emotional, and economic burden associated with advanced breast cancer.

Molecular Biomarkers in Breast Cancer

In recent years, the identification and analysis of molecular biomarkers have become crucial in understanding the pathogenesis, progression, and treatment of breast cancer. Molecular biomarkers provide valuable information about tumor biology, prognosis, and therapeutic response, thereby enabling a more personalized approach to patient management.

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The most well-established biomarkers in breast cancer are **estrogen receptor (ER)**, **progesterone receptor (PR)**, and **human epidermal growth factor receptor 2 (HER2)**. Based on their expression patterns, breast cancer is classified into four major molecular subtypes:

- **Luminal A (ER+/PR+, HER2−, low Ki-67)** — characterized by slow growth and favorable prognosis.
- **Luminal B (ER+/PR+, HER2+/−, high Ki-67)** — associated with more aggressive behavior and poorer outcomes than Luminal A.
- **HER2-enriched (ER−/PR−, HER2+)** — tends to grow faster but responds well to targeted therapy such as trastuzumab.
- **Triple-negative breast cancer (TNBC; ER−, PR−, HER2−)** — lacks common therapeutic targets and is associated with an unfavorable prognosis.

Mutations in the **BRCA1** and **BRCA2** genes play a significant role in hereditary breast cancer, accounting for approximately 5–10% of all cases. Carriers of these mutations have a markedly increased lifetime risk of developing breast and ovarian cancers. Genetic testing for BRCA mutations allows early preventive interventions, including prophylactic surgery and targeted therapy with **PARP inhibitors**.

Other emerging biomarkers, such as **PIK3CA**, **TP53**, **PTEN**, and **CDH1** mutations, as well as gene expression profiles (e.g., Oncotype DX, MammaPrint), contribute to predicting treatment response and disease recurrence. These molecular insights have transformed breast cancer from a uniform disease into a biologically diverse group of disorders, each requiring specific therapeutic strategies. Incorporating biomarker assessment into clinical practice allows oncologists to tailor treatment plans precisely, improving efficacy while minimizing unnecessary toxicity. Thus, molecular diagnostics play a pivotal role in modern oncology, bridging the gap between basic research and individualized patient care.

Targeted and Personalized Therapy Approaches

The advent of targeted and personalized therapy has revolutionized the treatment of breast cancer, shifting the paradigm from conventional “one-size-fits-all” approaches toward precision medicine based on individual tumor biology.

Targeted therapy focuses on specific molecular pathways that drive tumor growth and survival. The most successful example is the development of **HER2-targeted drugs**, such as *trastuzumab*, *pertuzumab*, *lapatinib*, and *ado-trastuzumab emtansine (T-DM1)*, which have dramatically improved survival outcomes for patients with HER2-positive breast cancer. Combination regimens involving these agents and chemotherapy provide superior response rates while reducing systemic toxicity.

For patients with **hormone receptor–positive (ER/PR+)** breast cancer, **endocrine therapy** remains the cornerstone of treatment. Agents such as *tamoxifen*, *aromatase inhibitors* (letrozole, anastrozole, exemestane), and *fulvestrant* inhibit estrogen-driven proliferation, thereby reducing recurrence risk. The integration of **CDK4/6 inhibitors** (palbociclib, ribociclib, abemaciclib) with endocrine therapy has further enhanced progression-free survival in metastatic settings.

In **triple-negative breast cancer (TNBC)**, where conventional targeted receptors are absent, progress has been achieved through **immunotherapy** and **PARP inhibitors**. Immune checkpoint

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inhibitors, including *pembrolizumab* and *atezolizumab*, have shown promising results, particularly in PD-L1–positive cases. Meanwhile, PARP inhibitors (*olaparib*, *talazoparib*) have become a standard option for patients with BRCA1/2 mutations, exploiting synthetic lethality mechanisms to induce tumor cell death.

Advances in **genomic profiling** and **next-generation sequencing (NGS)** allow for the identification of actionable mutations, enabling the development of individualized treatment regimens. Liquid biopsy and circulating tumor DNA (ctDNA) monitoring further support real-time assessment of treatment efficacy and disease progression.

Thus, targeted and personalized therapy represents a transformative step in breast cancer management. By tailoring treatment to the unique genetic and molecular profile of each patient, clinicians can achieve greater therapeutic precision, improved survival rates, and better quality of life.

Discussion

The integration of early detection programs with molecular biomarker assessment represents a milestone in modern oncology. Early diagnosis allows timely intervention, while molecular profiling ensures that patients receive the most effective and least toxic therapy tailored to their tumor's biology. Together, these approaches form the foundation of precision medicine in breast cancer.

Screening methods such as mammography, ultrasound, and MRI have proven their effectiveness in identifying early-stage lesions, thereby reducing mortality. However, disparities in access to these technologies remain a challenge, particularly in low- and middle-income countries. Expanding population-based screening programs, increasing public awareness, and improving access to diagnostic imaging are essential steps toward achieving equitable healthcare outcomes.

At the molecular level, the identification of biomarkers such as **ER, PR, HER2, and Ki-67** has provided a framework for classification, prognosis, and therapeutic decision-making. Advances in genomic analysis have expanded this framework, revealing complex molecular heterogeneity within breast cancer. For instance, even among patients with the same clinical stage, outcomes can vary significantly due to differences in gene expression and mutational status.

The success of **targeted therapies** such as *trastuzumab* for HER2-positive cases and *PARP inhibitors* for BRCA-mutated cancers highlights the clinical importance of biomarker-driven treatment. Furthermore, combining molecular diagnostics with real-time monitoring technologies such as liquid biopsy can improve treatment precision by detecting emerging resistance mutations or early signs of relapse.

Despite these advances, challenges remain. Not all patients benefit equally from current targeted therapies, and tumor heterogeneity often leads to resistance. Ongoing research into novel targets—such as PI3K/AKT/mTOR, FGFR, and immune checkpoints—holds promise for the next generation of therapeutic interventions. Additionally, integrating artificial intelligence (AI) and bioinformatics into diagnostic workflows can enhance accuracy, speed, and personalization in clinical decision-making.

In conclusion, the effective combination of early detection and molecular profiling transforms breast cancer care from a reactive to a proactive model. By addressing both biological complexity and

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healthcare accessibility, the global oncology community moves closer to achieving personalized, equitable, and curative management for all breast cancer patients.

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