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BREAST CANCER: EPIDEMIOLOGY, MOLECULAR CHARACTERISTICS, AND GENETIC BASIS (LITERATURE REVIEW)

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Abstract: Breast cancer is one of the most prevalent oncological diseases among women worldwide and is characterized by a complex interplay of epidemiological, biological, and genetic factors. This literature review analyzes the global and regional epidemiology of breast cancer, age-related patterns, molecular subtypes, and key genetic determinants associated with hereditary susceptibility. Particular emphasis is placed on the role of pathogenic variants in the BRCA1 and BRCA2 genes in disease pathogenesis, clinical risk assessment, and therapeutic decision-making. Furthermore, the clinical utility of next-generation sequencing (NGS) technologies in precision medicine is discussed, highlighting their applications in diagnosis, prognosis, treatment selection, and patient monitoring. The findings of this review underscore that the integration of population genomics and advanced molecular diagnostics into clinical practice has the potential to significantly reduce breast cancer–related morbidity and mortality.

Keywords: breast cancer, BRCA1, BRCA2, next-generation sequencing (NGS), precision medicine, molecular diagnostics

Introduction

Breast cancer (BC) represents one of the most frequently diagnosed malignancies among women globally, with a steadily increasing incidence and mortality rate observed over the past decades [1–3]. The high prevalence and clinically heterogeneous nature of breast cancer make it not only a major medical concern but also a significant socioeconomic challenge.

Global epidemiology

Breast cancer accounts for a substantial proportion of all malignancies diagnosed in women and was reported as the most commonly diagnosed cancer worldwide in 2020 [4]. The disease burden is rising particularly rapidly in countries undergoing economic and social transitions.

According to global estimates, more than 2.26 million new cases of breast cancer were diagnosed worldwide in 2020, accounting for approximately 12% of all newly diagnosed cancers [5]. In European countries, nearly 9% of women are estimated to develop breast cancer during their lifetime [6]. Although the disease is most frequently diagnosed after the age of 60, approximately 5–10% of cases occur in women younger than 40 years [7].

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Regional and age-related disparities

Significant differences in age-specific incidence patterns of breast cancer have been observed between Asian and Western populations. In many Asian countries, the peak incidence occurs between 40 and 50 years of age, whereas in Western countries it is more commonly observed between 60 and 70 years [8].

In developing regions, including parts of Africa, South America, and Asia, the incidence of breast cancer is increasing markedly. These trends are largely attributed to lifestyle changes, rising obesity rates, altered reproductive patterns, and increased life expectancy [9].

Breast cancer in Uzbekistan

In recent years, an increasing incidence of oncological diseases, particularly breast cancer, has also been observed in the Republic of Uzbekistan [10]. According to data from the International Agency for Research on Cancer (IARC), the age-standardized incidence rate of cancer in the country is 108.1 per 100,000 population [11]. Breast cancer remains the most frequently diagnosed malignancy among women in Uzbekistan.

Molecular and biological characteristics

Breast cancer is a highly heterogeneous disease in terms of etiology, histopathology, and molecular profile [12]. Contemporary molecular classification systems categorize breast cancer into several major subtypes, including Luminal A, Luminal B, HER2-positive, and triple-negative breast cancer (TNBC) [13]. Each subtype is associated with distinct biological behavior, clinical outcomes, and responses to therapy.

Genetic factors and hereditary predisposition (focus on BRCA)

Approximately 5–10% of breast cancer cases are attributed to inherited genetic alterations, with BRCA1 and BRCA2 genes playing a pivotal role due to their involvement in DNA damage repair pathways [14]. Pathogenic variants in these genes disrupt homologous recombination repair mechanisms, leading to genomic instability and a substantially increased risk of malignant transformation [15]. Identification of BRCA mutations is critical not only for cancer risk assessment but also for therapeutic decision-making, particularly regarding the use of PARP inhibitors.

NGS and modern approaches in molecular diagnostics

Recent advances in omics technologies have enabled the implementation of more precise and individualized approaches in precision medicine [16]. In this context, next-generation sequencing (NGS) technologies have assumed a central role, facilitating comprehensive analysis of the molecular landscape of breast cancer.

NGS is widely applied in breast cancer for diagnostic confirmation, prognostic stratification, therapeutic selection, and longitudinal patient monitoring [17]. Notably, NGS demonstrates high sensitivity and throughput for detecting pathogenic and likely pathogenic variants in BRCA1/2

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genes, making it an essential diagnostic tool for hereditary breast cancer syndromes. Moreover, NGS enables the identification of novel and population-specific variants, which is particularly relevant for underrepresented populations.

Discussion

The findings of this literature review highlight breast cancer as a multifactorial disease with significant global prevalence and pronounced molecular complexity. Epidemiological variations, particularly age- and region-specific differences, reflect the interplay between genetic susceptibility and environmental influences. Inherited mutations in BRCA1 and BRCA2 constitute a major molecular basis of breast cancer predisposition and have direct implications for clinical management.

The integration of NGS technologies into clinical practice has revolutionized breast cancer diagnostics and management. NGS facilitates a more comprehensive assessment of tumor heterogeneity, enables the discovery of novel genetic variants, and supports the development of personalized therapeutic strategies. In developing countries, including Uzbekistan, identifying population-specific genetic variants and evaluating their clinical significance represent critical steps toward implementing effective precision oncology frameworks.

Conclusion

Breast cancer remains a major global health challenge characterized by high incidence, molecular heterogeneity, and diverse clinical behavior. Integrating epidemiological data with molecular profiling, identifying hereditary alterations in BRCA genes, and expanding the clinical application of NGS technologies are essential for early detection, individualized treatment, and improved prognostic outcomes. Future population-based genomic studies and precision medicine–driven approaches are expected to play a crucial role in reducing breast cancer–associated morbidity and mortality.

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