NEW DIRECTIONS IN BREAST CANCER RESEARCH RELATED TO THE CHEK2 GENE: BIBLIOMETRIC AND MOLECULAR ANALYSIS.

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https://doi.org/10.5281/zenodo.17452980

ARTICLE INFO

Received: 23rd October 2025
Accepted: 24th October 2025
Online: 25th October 2025

KEYWORDS

CHEK2 gene, breast cancer, bibliometric analysis, pathogenic variant, DNA repair pathways, HRD (homologous recombination deficiency), intronic mutation, multi-organ cancer risk.

ABSTRACT

Breast cancer (BC) remains one of the most prevalent oncological diseases among women worldwide and continues to be a leading cause of female mortality. The CHEK2 gene, which plays a crucial role in DNA repair mechanisms, is of significant importance in studying hereditary risk factors contributing to cancer development. In this study, a bibliometric analysis was conducted based on 550 scientific articles published in the Scopus database between 2018 and 2022, and the data were updated with the most recent scientific sources published in 2024.

Introduction: The analysis revealed an exponential increase in the number of scientific publications related to the *CHEK2* gene. The most active research institutions were the Memorial Sloan Kettering Cancer Center and Harvard Medical School (USA), with F.J. Couch and M.K. Schmidt identified as leading authors in the field. The majority of articles were published in high-impact journals such as *Cancers*, *International Journal of Molecular Sciences*, and *Breast Cancer Research and Treatment*. Molecular studies published in 2024 have provided deeper insights into the pathogenic mechanisms of *CHEK2* gene variants in cancer development. Notably, it was found that *CHEK2* mutations are not associated with homologous recombination deficiency (HRD), which explains the limited efficacy of PARP inhibitors in patients carrying such variants [1]. In addition, a novel deep intronic variant, c.1009-118_1009-87delinsC, was characterized in 2024. This alteration was found to disrupt the pre-mRNA splicing process and is associated with an increased hereditary risk of breast cancer [2].

Furthermore, population-based analyses conducted in 2024 demonstrated that carriers of the *CHEK2* (c.1100delC) variant have an increased risk of developing cancers not only in the mammary glands but also in other organs. [3,4]. From a clinical perspective, such studies emphasize the need to individualize therapeutic approaches for *CHEK2*-associated cancer cases. Other investigations have confirmed that the efficacy of PARP inhibitors is primarily limited to conditions involving disruptions in HR-DDR pathways (e.g., *BRCA1/2* mutations), whereas in the case of *CHEK2*, the therapeutic response appears to be minimal or uncertain. [5,6].

Conclusion: Bibliometric and molecular analyses indicate that research on the *CHEK2* gene in 2024 has expanded not only in quantity but also in scope. Scientific attention is increasingly focused on the functional significance of genetic variants, their clinical impact, and the associated multi-organ cancer risks. These developments reinforce the role of the *CHEK2* gene as an important biomarker in future diagnostic strategies, genetic screening programs, and personalized cancer therapy systems..

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