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CRISPR-Based Molecular Diagnostics for Early Mutation Detection in High-Risk Breast and Ovarian Cancer Patients

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Abstract

Hereditary breast and ovarian cancers pose significant health challenges due to their aggressive nature and the high mortality associated with late-stage diagnosis. Early detection of pathogenic mutations in high-risk individuals, particularly in genes such as BRCA1 and BRCA2, is critical for improving patient outcomes through preventive and personalized interventions. Traditional genetic testing methods, while effective, face limitations related to cost, turnaround time, and accessibility. The emergence of CRISPR-based molecular diagnostics has introduced a novel, highly sensitive, and rapid approach for mutation detection. Utilizing programmable guide RNAs and the collateral cleavage activity of Cas enzymes, CRISPR diagnostics enable precise identification of cancer-associated mutations even at low allele frequencies in liquid biopsy samples. This review explores the advancements, advantages, and challenges of CRISPR-based molecular diagnostic platforms in the context of early mutation detection for breast and ovarian cancer patients at high genetic risk. It also discusses future prospects, including integration with microfluidics and artificial intelligence, to enhance clinical application and accessibility worldwide.

Keywords: CRISPR, Molecular diagnostics; Early mutation detection; Breast cancer; Ovarian cancer; BRCA1; BRCA2; Liquid biopsy; High-risk patients; Genetic testing

Introduction

Breast and ovarian cancers remain among the most prevalent and deadly malignancies affecting women worldwide [1]. According to the World Health Organization (WHO), breast cancer is the most commonly diagnosed cancer and the leading cause of cancer-related deaths in women globally [2]. Ovarian cancer, though less common, is notorious for its late-stage diagnosis and poor prognosis. Early detection of genetic mutations in high-risk individuals can significantly improve outcomes by enabling preventive strategies, targeted surveillance, and personalized therapies [3]. The advent of CRISPR (Clustered Regularly Interspaced Short Palindromic Repeats) technology has revolutionized molecular diagnostics, offering unprecedented precision, sensitivity, and speed in detecting genetic alterations [4]. This article explores the role of CRISPR-based molecular diagnostics in early mutation detection for high-risk breast and ovarian cancer patients, emphasizing its clinical significance, technological advances, and future potential [5].

Hereditary breast and ovarian cancers are frequently linked to germline mutations in tumor suppressor genes such as BRCA1 and BRCA2. Women carrying these mutations face a significantly elevated lifetime risk of developing breast and ovarian cancers compared to the general population [6]. Other genes, including TP53, PALB2, RAD51C, and CHEK2, also contribute to hereditary cancer susceptibility. Traditional genetic testing methods, including Sanger sequencing and next-generation sequencing (NGS), have been instrumental in identifying mutations in these genes [7]. However, these approaches are often expensive, time-consuming, and require sophisticated infrastructure and bioinformatics expertise, limiting their accessibility, especially in low-resource settings. Moreover, the need for rapid, cost-effective, and highly sensitive diagnostic tools to screen large populations of high-risk individuals has become increasingly urgent [8].

CRISPR Technology: A new era in molecular diagnostics

CRISPR-Cas systems, originally discovered as an adaptive immune mechanism in bacteria, have been repurposed for genome editing and molecular diagnostics. The CRISPR-Cas9 nuclease enables targeted DNA cleavage, while other Cas proteins, such as Cas12 and Cas13, have been adapted for detecting nucleic acid sequences with high specificity. CRISPR-based diagnostics leverage the programmable nature of guide RNAs (gRNAs) to recognize specific DNA or RNA sequences associated with cancer-related mutations. Upon target recognition, the Cas enzymes exhibit collateral cleavage activity that can be harnessed to produce detectable signals, often through fluorescence or colorimetric readouts. This mechanism underpins platforms such as SHERLOCK (Specific High Sensitivity Enzymatic Reporter Unlocking) and DETECTR (DNA Endonuclease Targeted CRISPR Trans Reporter), which have demonstrated remarkable sensitivity and specificity in detecting low-abundance mutations.

CRISPR diagnostics can detect single nucleotide polymorphisms (SNPs) and small insertions or deletions (indels) with exceptional precision. This capability is crucial for identifying pathogenic mutations in BRCA1/2 genes and other cancer predisposition loci, even when present at low allele frequencies in circulating tumor DNA (ctDNA) or cell-free DNA (cfDNA) from liquid biopsies.

Rapid and cost-effective testing

Traditional sequencing methods can take days to weeks to return results, delaying clinical decision-making. CRISPR-based assays

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can deliver results within hours, allowing for timely interventions. Additionally, the simplicity of CRISPR diagnostics reduces reliance on expensive instrumentation and specialized personnel, making them suitable for point-of-care testing and wider population screening. Liquid biopsy techniques enable the collection of blood or urine samples to detect ctDNA, avoiding invasive tissue biopsies. CRISPR diagnostics' high sensitivity enhances the utility of liquid biopsies for early mutation detection, facilitating frequent monitoring of high-risk patients and early identification of cancer onset. Several recent studies underscore the potential of CRISPR diagnostics in hereditary cancer mutation detection:

Researchers have developed CRISPR-Cas12-based assays capable of identifying common BRCA1 and BRCA2 mutations from patient plasma samples. These assays demonstrated a limit of detection as low as 0.1% mutant allele frequency, outperforming some conventional PCR-based methods. Using combinatorial gRNAs, CRISPR platforms can simultaneously detect multiple mutations in a single reaction, facilitating comprehensive genetic screening for high-risk patients.

The coupling of CRISPR diagnostics with microfluidic devices enables automated sample processing and real-time detection, paving the way for portable, user-friendly diagnostic kits suitable for clinical and field settings.

Ethical and privacy concerns, genetic testing raises ethical questions regarding data privacy, informed consent, and potential discrimination. Appropriate counseling and data protection frameworks are essential.

Conclusion

CRISPR-based molecular diagnostics represent a transformative advancement in the early detection of mutations associated with high-risk breast and ovarian cancers. Their exceptional sensitivity, rapid turnaround time, and adaptability for non-invasive testing position them as powerful tools in personalized medicine. While challenges related to specificity, regulation, and ethics must be addressed, ongoing research and technological innovations promise to refine and expand their clinical utility. By enabling early mutation detection, CRISPR diagnostics can facilitate preventive interventions, improve patient outcomes, and ultimately reduce the global burden of hereditary breast and ovarian cancers.

References

1. Parham G (2010) cervical cancer prevention in HIV-infected women in resource-limited settings. HIV Therapy 4: 625-628.
2. Arbyn M, Weiderpass E, Bruni L, De Sanjose S, Saraiya M, et al. (2020) Estimates of incidence and mortality of cervical cancer in 2018: a worldwide analysis. THE LANCET-Global Health 8: 191-203.
3. Fernandez E (2020) Climate Change Will Give Rise to More Cancers. UCSF Research Journal.
4. Smith JS, Green J, Berrington de Gonzalez A, Appleby P, Peto J, Plummer M, et al (2003) Cervical cancer and use of hormonal contraceptives: a systematic review. Lancet 361: 1159-67.
5. Rabkin CS, Biggar RJ, Baptiste MS, Abe T, Kohler BA, et al. (1993) Cancer incidence trends in women at high risk of human immunodeficiency virus (HIV) infection. Int J Cancer 55:208-12.
6. Mapanga W, Brown GB, Singh E (2019) Knowledge, attitudes and practices of young people in Zimbabwe on cervical cancer and HPV, current screening methods and vaccination. BMC cancer 19: 843.
7. Coutinho RA (2000) highly active antiretroviral therapy and incidence of cancer in human immunodeficiency virus-infected adults. J Natl Cancer Inst 92: 1823.
8. Frisch M, Biggar RJ, Goedert JJ (2000) Human papillomavirus-associated cancers in patients with human immunodeficiency virus infection and acquired immunodeficiency syndrome. J Natl Cancer Inst 92: 1500-1510.