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LETTER TO THE EDITOR Open Access

From sensitivity to public health: integrating cfMeDIP-seq into early breast cancer detection strategies

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To the Editor:

I had the pleasure of reading the article [1] by Grisolia et al., titled "Differential methylation of circulating free DNA assessed through cfMeDiP as a new tool for breast cancer diagnosis and detection of BRCA1/2 mutation," published in the *Journal of Translational Medicine*. I am impressed by the potential of this study in early breast cancer diagnosis and personalized treatment. The study demonstrates the application of cfMeDIP-seq technology in identifying methylation patterns of circulating free DNA (cfDNA) in patients with BRCA1/2 mutations, providing strong evidence for early detection and individualized risk assessment through non-invasive methods. I would like to offer a few suggestions to further enhance the clinical and public health significance of this research.

Potential for early detection and primary prevention

The cfDNA methylation-based detection method proposed in the article provides a new approach for the early diagnosis of breast cancer in BRCA1/2 mutation carriers, contributing to primary prevention. Early identification of high-risk individuals allows for interventions before the onset of clinical symptoms, such as regular monitoring, lifestyle adjustments, and pharmacological prevention (e.g., selective estrogen receptor modulators). However, the current research primarily focuses on the technical sensitivity and specificity of the method. We suggest that future studies further explore how this detection method can be integrated with community and public health screening programs to optimize early screening and intervention strategies for high-risk populations [2].

Risk assessment and health management

Carriers of BRCA1/2 mutations face a significantly elevated risk of breast cancer. By analyzing methylation status, cfMeDIP-seq technology provides a means for personalized risk assessment. However, the genetic risk associated with mutations can be abstract to the general population, often leading to anxiety. Therefore, it is important to develop personalized risk communication strategies that help high-risk individuals understand their test results and adopt proactive health measures. These risk assessment tools should also be integrated into comprehensive health management plans, including individualized exercise, dietary guidance, and psychological support, to help reduce the incidence of breast cancer [3].

Cost-effectiveness and feasibility of genetic testing

While the article highlights the potential of cfMeDIP-seq technology for early breast cancer detection, its high cost and reliance on specialized equipment may limit its application in large-scale screening programs. Preventive

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medicine emphasizes the importance of cost-effective approaches to disease prevention and health promotion. We recommend that future research not only focus on the diagnostic efficacy of this technology but also include cost-effectiveness analyses to assess its feasibility in broader screening settings. Additionally, exploring the combination of this technique with other, more affordable screening methods, such as mammography and ultrasound, could lead to more effective and accessible early detection strategies [4].

Multidisciplinary collaboration and secondary prevention

Early identification of BRCA1/2 mutation carriers facilitates targeted secondary prevention, including early diagnosis and timely treatment. Multidisciplinary collaboration is essential, involving oncologists, genetic counselors, preventive medicine specialists, and mental health professionals to provide comprehensive care. For example, a holistic health management plan for high-risk individuals identified through BRCA testing could include regular imaging, pharmacological prevention, lifestyle interventions, and psychological support, aiming to delay or prevent the onset of breast cancer.

Long-term follow-up and tertiary prevention

Although the study mainly focuses on the use of cfDNA methylation analysis for early diagnosis, its potential role in tertiary prevention is equally noteworthy. For patients already diagnosed with breast cancer, this detection method could be used to monitor residual disease or assess treatment responses, thereby optimizing therapeutic strategies. We advocate for the establishment of long-term follow-up mechanisms for high-risk individuals and treated patients to enable continuous health monitoring, detect recurrence or metastasis early, and improve long-term outcomes [5].

Conclusion

The research by Grisolia et al. offers new perspectives and methods for early breast cancer screening and risk assessment in BRCA1/2 mutation carriers, with significant clinical application potential. By advancing the use of new technologies in high-risk population screening, promoting personalized health management, enhancing multidisciplinary collaboration, and addressing cost-effectiveness, this research could further increase its public health value. We look forward to future studies that make progress in these areas, providing stronger support for the prevention of breast cancer and reducing the burden of the disease on patients.

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