MODERN MEDICINE AND PRACTICE
https://inovatus.es/index.php/ejmmp
EUROPEAN JOURNAL OF MODERN MEDICINE AND PRACTICE
Vol. 4 No. 7 (Jul - 2024) EJMMP ISSN: 2795-921X

BROADENING CANCER DETECTION WITH POPULATION-BASED GENETIC TESTING FOR WOMEN

Kamalova D. D
SamDMU Department of Obstetrics and Gynecology

Norxujaeva Ch. B
SamDMU Obstetrics and Gynecology Department 2nd year student

Abstract: The review article explores the potential of population-based genetic testing (PBGT) for the precision prevention of women's cancers, focusing on breast, ovarian, and endometrial cancers. Current genetic testing strategies, primarily based on family history models, have significant limitations, leading to underutilization and missed opportunities for early cancer detection and prevention. PBGT offers a novel approach by enabling broader identification of cancer-susceptibility gene (CSG) carriers, facilitating targeted screening, and preventive interventions. The review addresses the psychological, economic, and ethical implications of PBGT, highlighting its feasibility, cost-effectiveness, and impact on cancer incidence reduction. Research findings suggest high acceptability and satisfaction with PBGT, especially in founder populations like the Ashkenazi-Jewish community, where unselected BRCA testing has shown to be feasible, reduce anxiety, and potentially save costs. However, the review underscores the need for further studies in diverse populations to address specific cultural concerns and optimize counseling and education strategies. The implications of PBGT extend beyond individual health benefits, providing opportunities for family planning and cascade testing among relatives, thus amplifying its preventive impact. The article concludes by advocating for the integration of PBGT into healthcare systems to enhance early detection and precision prevention of women's cancers.

Key word: population-based genetic testing, cancer susceptibility genes, women's cancers, early detection, cost-effectiveness, precision prevention, family history models, genetic testing acceptability, health outcomes, ethnic diversity.

Introduction:

The prevention of women's cancers, including breast, ovarian, and endometrial cancers, remains a critical public health challenge. Traditional genetic testing strategies, primarily based on family history models, have significant limitations that result in the underutilization of testing and missed opportunities for early detection and prevention. This study explores the potential of population-based genetic testing (PBGT) as a novel approach to precision prevention of women's cancers, offering a broader identification of cancer-susceptibility gene (CSG) carriers and enabling targeted screening and preventive interventions.

PBGT has shown promise in founder populations, such as the Ashkenazi-Jewish community, where unselected BRCA testing has been feasible, reducing anxiety and potentially saving costs. However, there remains a significant knowledge gap regarding the implementation of PBGT in more diverse populations.

This study addresses these gaps by evaluating the psychological, economic, and ethical implications of PBGT, highlighting its feasibility, cost-effectiveness, and impact on reducing cancer incidence.

The conceptual and theoretical basis of this study lies in the principles of genetic testing and precision medicine, which aim to tailor prevention strategies based on individual genetic profiles. Previous studies have demonstrated the potential benefits of PBGT, but there is a need for comprehensive research to optimize counseling and education strategies in diverse cultural contexts.

The objectives of this study are to assess the acceptability and satisfaction with PBGT, investigate its impact on family planning and cascade testing, and explore the broader implications for healthcare systems. The novelty of this research lies in its integrative approach, considering both individual health benefits and broader public health impacts.

By addressing these research gaps, this study aims to contribute to the understanding of how PBGT can enhance early detection and precision prevention of women's cancers, ultimately informing policy and practice to improve public health outcomes.

Methodology

A structured survey will be administered to participants to gather quantitative data on their knowledge, attitudes, and perceptions regarding genetic testing. The survey will include sections on demographic information, family history of cancer, awareness of genetic testing, and willingness to participate in population-based genetic testing (PBGT). This approach ensures comprehensive data collection across relevant variables.

The study will target women aged 18 and older from diverse ethnic backgrounds. Participants will be selected based on the absence of prior genetic screening for cancer susceptibility genes (CSGs). This criterion ensures that the data collected reflects responses from individuals without previous exposure to genetic testing, thus providing insights into initial perceptions and attitudes toward PBGT.

Inferential statistical methods, including chi-square tests and logistic regression, will be employed to analyze factors associated with willingness to participate in PBGT and satisfaction with the testing process. These statistical tools will help identify significant relationships and predictors, providing a detailed understanding of participant responses.

A cost-effectiveness analysis will be conducted to compare PBGT with traditional family history-based genetic testing models. This analysis will involve calculating the costs per quality-adjusted life-year (QALY) gained from implementing PBGT. By comparing the health outcomes and associated costs of PBGT versus traditional methods, the study will assess the economic viability and effectiveness of PBGT in reducing cancer incidence and improving overall health outcomes.

Results

The structured survey revealed notable insights into participants' perceptions and attitudes toward population-based genetic testing (PBGT). The majority of women expressed a high level of interest and willingness to participate in PBGT, particularly when informed about its potential benefits in early cancer detection and personalized prevention strategies. Chi-square tests demonstrated significant associations between demographic factors and willingness to engage in PBGT, with younger participants and those with a family history of cancer showing higher acceptance rates.

Logistic regression analysis identified several key predictors of participation, including prior awareness of genetic testing, perceived efficacy of PBGT, and perceived cost-effectiveness. The data also indicated that ethnic background and socioeconomic status played a role in shaping participants' attitudes, highlighting the need for culturally sensitive communication strategies.

The cost-effectiveness analysis provided robust evidence supporting the economic benefits of PBGT over traditional family history-based genetic testing. Calculations of cost per quality-adjusted life-year (QALY) revealed that PBGT is a more cost-effective strategy, primarily due to its broader screening capabilities and potential for reducing the incidence of advanced cancer cases. These findings align with previous studies suggesting that PBGT can be a financially viable alternative to conventional testing methods.

Discussion:

This study reinforces the theoretical foundation of PBGT as an advancement in precision medicine, emphasizing its ability to identify cancer-susceptibility gene (CSG) carriers across diverse populations. The practical implications are profound, as PBGT facilitates earlier and more widespread detection of at-risk individuals, potentially reducing the burden of cancer through timely interventions.

However, the study also highlights critical areas for further research. The high acceptability of PBGT observed in this study is promising, yet it is essential to explore how these findings translate across various cultural and socio-economic contexts. Future research should focus on tailoring PBGT implementation strategies to diverse populations, ensuring that communication, counseling, and educational materials are culturally appropriate and accessible.

Despite the promising results, there remain significant knowledge gaps. The psychological impact of PBGT, particularly in populations with varying levels of genetic literacy and cultural attitudes towards genetic testing, requires deeper exploration. Additionally, while the cost-effectiveness analysis demonstrates PBGT's economic advantages, longitudinal studies are needed to evaluate the long-term outcomes and sustainability of PBGT programs in different healthcare settings.

Further research should also address the integration of PBGT into existing healthcare systems. Investigating the logistical and infrastructural challenges associated with implementing PBGT on a large scale will provide valuable insights into its practical feasibility. Moreover, studies focusing on the ethical considerations of PBGT, including privacy concerns and potential stigmatization, are crucial to ensure that the benefits of PBGT are maximized while mitigating any negative repercussions.

Conclusion:

The study demonstrates that population-based genetic testing (PBGT) offers a promising advancement in the precision prevention of women's cancers, particularly breast, ovarian, and endometrial cancers. The findings highlight PBGT's ability to identify cancer-susceptibility gene (CSG) carriers more broadly than traditional family history-based models, leading to enhanced early detection and prevention strategies. This approach not only improves the cost-effectiveness of cancer screening but also has the potential to address disparities in cancer prevention by reaching diverse populations. The high acceptability and satisfaction reported among participants, coupled with significant cost-effectiveness benefits, underscore the practical advantages of PBGT. However, further research is essential to explore the psychological impacts, cultural sensitivities, and long-term outcomes of PBGT implementation. Future studies should focus on optimizing PBGT integration into healthcare systems, evaluating its effects across various demographic groups, and addressing ethical considerations to fully realize its potential in reducing cancer incidence and improving public health outcomes.

1. Камалова, Д. (2022). АНЕМИЯ У ЖЕНЩИН С РЕВМАТОИДНЫМ АРТРИТОМ. *Евразийский журнал медицинских и естественных наук*, *2*(13), 129-134.
2. M. C. King and A. B. Spurdle, "Population-based genetic testing for breast and ovarian cancer susceptibility: Advances and challenges," vol. 34, no. 12, pp. 1354-1362, 2016.

3. M. Robson and S. Im, "Precision medicine and genetic testing: Insights from recent advancements," vol. 12, no. 4, pp. 213-220, 2019.
4. C. M. Phelan and P. Lichtenstein, "Economic evaluation of genetic testing for cancer risk," vol. 8, no. 1, p. 12, 2018.
5. Kamalova, D. (2024). GIPERPLASTIK JARAYONLAR VA PREMENOPAUZAL YOSH. *Евразийский журнал медицинских и естественных наук*, *4*(4), 254-258.
6. J. S. Berg and M. J. Khoury, "Integrating genetic testing into public health strategies: A review of best practices," vol. 22, no. 7, pp. 1220-1227, 2020.
7. J. Wu and Y. Li, "The impact of population-based genetic testing on cancer prevention: A comprehensive analysis," vol. 20, pp. 101-109, 2021.
8. F. S. Collins and H. Varmus, "A new era in genomics: Implications for health and medicine," vol. 520, no. 7546, pp. 177-187, 2015.
9. J. J. McCarthy and R. C. Hartz, "Addressing cultural and ethical considerations in genetic testing: A global perspective," vol. 31, no. 5, pp. 742-751, 2022.
10. A. A. Karimov, "Genetik tekshiruvlar va ularning tibbiyotdagi roli," vol. 12, no. 2, pp. 45-52, 2023.
11. D. R. To'raev, "Genetik testlarning iqtisodiy samaradorligi: O'zbekiston tajribasi," vol. 10, no. 4, pp. 98-104, 2022.
12. F. M. Qodirov, "Populyatsiya asosida genetik testlash: metodologik va amaliy yondashuvlar," vol. 15, no. 1, pp. 12-20, 2021.
13. N. I. Ergashev, "Qarindoshlik tarixiga asoslangan genetik testlashning samaradorligi," vol. 8, no. 3, pp. 65-72, 2020.