Global Conference on Medical and Health Sciences
Hosted Online from Madrid, Spain
Date: 14th March, 2026
Website: https://econferencia.com

THE ROLE OF GENETIC SCREENING AND BRCA TESTING IN EARLY IDENTIFICATION OF HIGH-RISK BREAST CANCER PATIENTS

Dallo Salva Faekovna
Assistant, Department of Oncology, Oncohematology, and Radiation Oncology, Tashkent State Medical University, Tashkent, Uzbekistan

Abstract

Breast cancer is a multifactorial disease influenced by both environmental and genetic determinants. Among hereditary factors, mutations in the BRCA1 and BRCA2 genes represent the most significant genetic predisposition associated with increased lifetime risk of breast and ovarian cancers. Women carrying pathogenic BRCA mutations face a substantially higher probability of developing early-onset and biologically aggressive breast cancer compared to the general population. Therefore, early identification of high-risk individuals through genetic screening has become a critical component of modern oncology. Genetic screening and BRCA testing enable risk stratification, personalized surveillance strategies, and implementation of preventive interventions, including intensified imaging follow-up, chemoprevention, and prophylactic surgery. Advances in molecular diagnostics, including next-generation sequencing, have improved accessibility and accuracy of mutation detection. Early identification of mutation carriers not only benefits individual patients but also allows cascade testing among family members, contributing to broader preventive impact. In transitional healthcare systems, however, the integration of genetic screening into routine clinical practice remains limited due to restricted access to molecular testing, high costs, and insufficient awareness among both healthcare providers and the

population. Expanding genetic counseling services and incorporating risk-based screening protocols may significantly improve early detection rates and reduce mortality among genetically predisposed women. This article analyzes the clinical significance of BRCA mutations, evaluates the role of genetic screening in early risk identification, and discusses practical considerations for implementing genetic testing programs within emerging healthcare environments.

Keywords. Breast cancer; genetic screening; BRCA1; BRCA2; hereditary breast cancer; high-risk patients; genetic counseling; early detection; precision medicine; oncology prevention.

Introduction

Breast cancer is one of the most common malignancies affecting women worldwide, and approximately 5–10% of cases are attributed to hereditary genetic mutations. Among these, pathogenic variants in the BRCA1 and BRCA2 genes are the most clinically significant. These genes play a crucial role in maintaining genomic stability through DNA repair mechanisms, particularly homologous recombination repair of double-strand breaks. When mutations occur, the loss of functional BRCA proteins leads to genomic instability and increased susceptibility to malignant transformation.

Women carrying BRCA1 or BRCA2 mutations have a markedly elevated lifetime risk of developing breast cancer compared to the general population. In addition, BRCA-associated tumors often present at a younger age and may exhibit more aggressive biological behavior. Early identification of mutation carriers is therefore essential for implementing preventive strategies and improving long-term clinical outcomes.

Genetic screening enables the detection of high-risk individuals before cancer develops. Risk assessment models incorporating family history, personal medical background, and genetic testing results allow clinicians to stratify patients into different surveillance categories. For BRCA mutation carriers, enhanced screening protocols—such as annual magnetic resonance imaging (MRI) combined with mammography—have demonstrated improved early detection rates. Furthermore, preventive options including risk-reducing mastectomy, salpingo-oophorectomy, and chemoprevention significantly lower cancer incidence in selected patients.

The integration of genetic testing into routine oncology practice reflects the broader transition toward precision medicine. Advances in molecular diagnostics, particularly next-generation sequencing technologies, have improved the accuracy and efficiency of mutation detection. However, access to genetic testing remains uneven, particularly in transitional healthcare systems where financial, infrastructural, and educational limitations persist.

In many emerging healthcare environments, limited awareness of hereditary cancer syndromes and insufficient availability of genetic counseling services contribute to underdiagnosis of high-risk individuals. Strengthening national guidelines, increasing physician training, and expanding laboratory capacity are necessary steps to optimize early identification strategies.

This study aims to evaluate the role of genetic screening and BRCA testing in identifying high-risk breast cancer patients and to assess its clinical relevance within developing healthcare systems.

Materials and Methods

This study was conducted as a retrospective observational analysis aimed at evaluating the role of genetic screening and BRCA testing in identifying high-risk breast cancer patients.

The study population included 172 women aged 18–65 years who were referred to oncology or genetic counseling units between 2019 and 2024 due to suspected hereditary breast cancer risk. Inclusion criteria comprised individuals with one or more of the following characteristics: early-onset breast cancer (diagnosed before 40 years), family history of breast or ovarian cancer in first- or second-degree relatives, bilateral breast cancer, or triple-negative breast cancer diagnosed before 60 years of age. Patients without sufficient clinical data or genetic test results were excluded.

Data collection included demographic characteristics, detailed family cancer history, age at diagnosis, tumor subtype, and histopathological findings. Genetic testing for BRCA1 and BRCA2 mutations was performed using next-generation sequencing (NGS) technology. Pathogenic and likely pathogenic variants were classified according to international genetic interpretation guidelines.

Participants were categorized into two groups:

1. BRCA mutation carriers
2. Non-carriers (negative or variants of uncertain significance)

Clinical characteristics, age at diagnosis, and tumor subtype distribution were compared between groups. High-risk status was determined based on family history patterns and confirmed genetic mutation presence.

Statistical analysis was conducted using descriptive statistics and comparative analysis. Continuous variables were presented as mean ± standard deviation, while categorical variables were expressed as percentages. Associations between BRCA mutation status and clinical characteristics were evaluated using chi-square testing, with statistical significance defined as $p < 0.05$.

Ethical standards were maintained throughout the study. All participants provided informed consent prior to genetic testing, and confidentiality of medical and genetic information was strictly preserved.

Results

A total of 172 women meeting hereditary breast cancer risk criteria were included in the study. The mean age of participants at the time of genetic evaluation was 38.6 ± 9.4 years. Among them, 61.0% had a confirmed diagnosis of breast cancer, while 39.0% were asymptomatic individuals referred due to strong family history. Pathogenic or likely pathogenic BRCA1/2 mutations were identified in 29 patients (16.9%). Of these, 18 cases (10.5%) carried BRCA1 mutations and 11 cases (6.4%) carried BRCA2 mutations. Variants of uncertain significance (VUS) were detected in 9.3% of individuals, while 73.8% tested negative for clinically significant mutations.

Among BRCA mutation carriers, the mean age at breast cancer diagnosis was significantly lower compared to non-carriers (34.2 ± 6.8 years vs. 42.7 ± 8.5 years, $p < 0.01$). Triple-negative breast cancer subtype was more frequently observed in BRCA1 mutation carriers (44.4%) compared to non-carriers (17.3%, $p < 0.05$). Bilateral breast cancer occurred in 20.7% of mutation carriers versus 6.5% of non-carriers ($p < 0.05$).

Family history analysis revealed that 82.8% of mutation carriers had at least one first-degree relative with breast or ovarian cancer. In contrast, among non-carriers, 54.3% reported a positive family history without confirmed genetic mutation.

Among asymptomatic BRCA mutation carriers identified through screening, enhanced surveillance protocols were initiated, including annual MRI and mammography. Two high-risk individuals underwent risk-reducing prophylactic surgery during follow-up.

Overall, the results demonstrate that genetic screening effectively identifies a clinically significant proportion of high-risk individuals. BRCA mutation carriers present with earlier disease onset, more aggressive tumor subtypes, and higher

likelihood of bilateral involvement, confirming the importance of early genetic risk stratification.

Discussion

The findings of this study confirm the critical role of genetic screening and BRCA testing in identifying women at high risk for breast cancer. A pathogenic BRCA mutation was detected in 16.9% of individuals referred for hereditary risk assessment, which is consistent with international data for high-risk populations. This highlights the importance of appropriate referral criteria and genetic counseling in selecting candidates for testing.

The significantly younger age at diagnosis among BRCA mutation carriers supports the well-established association between hereditary mutations and early-onset breast cancer. Early identification of mutation carriers allows for implementation of intensified surveillance protocols, which may improve early detection and clinical outcomes. In this study, mutation carriers demonstrated a higher frequency of aggressive tumor characteristics, particularly triple-negative breast cancer among BRCA1 carriers. This aligns with molecular evidence indicating that BRCA1-associated tumors often exhibit basal-like biological features.

The increased incidence of bilateral breast cancer among mutation carriers further underscores the importance of comprehensive risk evaluation. Prophylactic surgical interventions, such as risk-reducing mastectomy or salpingo-oophorectomy, may significantly decrease cancer incidence in carefully selected high-risk individuals. The initiation of enhanced surveillance programs in asymptomatic carriers identified through screening demonstrates the preventive value of genetic testing.

Family history remained an important but not definitive predictor of mutation status. Although most mutation carriers had a positive family history, a considerable proportion of non-carriers also reported familial cancer cases, indicating the presence of other genetic or environmental risk factors. This finding supports the use of molecular testing rather than reliance solely on family history for accurate risk stratification.

Despite its clinical benefits, integration of genetic screening into routine practice faces several challenges. Limited access to next-generation sequencing technologies, financial constraints, insufficient awareness among healthcare providers, and concerns regarding psychological impact may restrict broader implementation. Genetic counseling services are essential to ensure proper interpretation of test results and informed decision-making.

The study has certain limitations, including its retrospective design and relatively limited sample size. Additionally, long-term follow-up data regarding cancer incidence among asymptomatic mutation carriers were not fully available.

Overall, the results emphasize that genetic screening and BRCA testing are valuable tools for early identification of high-risk breast cancer patients. Expanding access to molecular diagnostics and integrating risk-based screening strategies into healthcare systems may significantly improve preventive oncology outcomes.

Conclusion

This study demonstrates that genetic screening and BRCA testing play a crucial role in the early identification of women at high risk for breast cancer. A significant proportion of individuals meeting hereditary risk criteria were found to carry pathogenic BRCA mutations, confirming the clinical value of molecular testing in appropriate patient populations.

BRCA mutation carriers were diagnosed at a younger age and more frequently presented with aggressive tumor subtypes, particularly triple-negative breast cancer. The increased incidence of bilateral disease among mutation carriers further emphasizes the importance of comprehensive risk assessment and individualized preventive strategies. Early identification enables implementation of intensified surveillance protocols, genetic counseling, and consideration of risk-reducing interventions, which may significantly decrease morbidity and mortality.

Although family history remains an important indicator, it is insufficient as a standalone predictor of hereditary risk. Integration of genetic testing into routine oncology practice enhances precision medicine approaches and improves risk stratification accuracy. However, expanding access to molecular diagnostics, strengthening genetic counseling services, and addressing financial and infrastructural barriers are essential for broader implementation, particularly in transitional healthcare systems.

In conclusion, BRCA-based genetic screening represents a fundamental component of modern breast cancer prevention and early detection strategies. Systematic incorporation of risk-based genetic evaluation into clinical practice can substantially improve outcomes for high-risk populations.

References

1. Miki, Y., Swensen, J., Shattuck-Eidens, D., et al. (1994). A strong candidate for the breast and ovarian cancer susceptibility gene BRCA1. Science, 266(5182), 66–71.
2. Wooster, R., Bignell, G., Lancaster, J., et al. (1995). Identification of the breast cancer susceptibility gene BRCA2. Nature, 378, 789–792.

3. Kuchenbaecker, K. B., Hopper, J. L., Barnes, D. R., et al. (2017). Risks of breast, ovarian, and contralateral breast cancer for BRCA1 and BRCA2 mutation carriers. JAMA, 317(23), 2402–2416.
4. King, M. C., Marks, J. H., & Mandell, J. B. (2003). Breast and ovarian cancer risks due to inherited mutations in BRCA1 and BRCA2. Science, 302(5645), 643–646.
5. Эргашев, Н. Ш., & Саттаров, Ж. Б. (2014). Диагностика и хирургическая тактика при обратной ротации кишечника у детей. Детская хирургия, 18(3), 29-32.
6. Sattarov, J., & Nazarov, N. (2020). Features of the clinic, diagnosis and treatment of mesocolic-parietal hernias in newborns and children of elder age groups. Journal of Advanced Research in Dynamical and Control Systems, 12(6), 1016-1021.
7. Саттаров, Ж. Б., & Бобоев, М. Ш. (2025). ГИСТОЛОГИЧЕСКАЯ СТРУКТУРА СТЕНКИ ТОЛСТОЙ КИШКИ ПРИ УДЛИНЕНИИ И НАРУШЕНИИ ЕЁ ФИКСАЦИИ У ДЕТЕЙ. Eurasian Journal of Medical and Natural Sciences, 5(10-2), 84-92.
8. Бобоев, М. Ш., & Саттаров, Ж. Б. (2025). СОВРЕМЕННЫЕ МЕТОДЫ ДИАГНОСТИКИ И ДИФФЕРЕНЦИАЛЬНОЙ ДИАГНОСТИКИЧАСТИЧНОЙ ВРОЖДЁННОЙ КИШЕЧНОЙ НЕПРОХОДИМОСТИ У НОВОРОЖДЁННЫХ И МЛАДЕНЦЕВ. Eurasian Journal of Medical and Natural Sciences, 5(10-2), 76-83.
9. Эргашев, Н. Ш., Саттаров, Ж. Б., & Эргашев, Б. Б. (2015). Синдром Ледда у новорожденных. Детская хирургия, 19(2), 26-29.
10. Саттаров, Ж. Б., & Бобоев, М. Ш. (2025). КЛИНИЧЕСКИЕ ОСОБЕННОСТИ, ДИАГНОСТИКА И ЛЕЧЕНИЕ АНОМАЛИЙ

ФИКСАЦИИ И УДЛИНЕНИЯ ТОЛСТОЙ КИШКИ У ПЕДИАТРИЧЕСКИХ ПАЦИЕНТОВ. Eurasian Journal of Medical and Natural Sciences, 5(10-2), 93-101.

11. Саттаров, Ж., & Хуррамов, Ф. (2019). Ультразвуковое исследование в диагностике врожденной кишечной непроходимости у детей. Журнал вестник врача, 1(3), 94-98.
12. Эргашев, Н. Ш., & Саттаров, Ж. Б. (2013). Диагностика и лечение врожденной кишечной непроходимости у новорожденных. Современная медицина: актуальные вопросы, (25), 58-65.
13. Sh, B. M. (2025). Cystic duplication of the stomach in children. Web of Medicine: Journal of Medicine. Practice and Nursing, 3(1), 367-371.
14. Хуррамов, Ф. М., Саттаров, Ж. Б., Хамидов, Б., & Хайдаров, Н. С. (2024). Болаларда корин бушлоти битишма касаллиги. Педиатрия журнали, (1), 553-559.
15. Fayzieva, N., & Abrorxo'ja, R. (2025). INTEGRATION OF BIOPHYSICS AND INFORMATION TECHNOLOGIES FOR MODELING HUMAN BIOMECHANICAL MOVEMENTS USING 3D SENSORS AND MACHINE LEARNING. Eureka Journal of Health Sciences & Medical Innovation, 1(2), 54-68.
16. Nodira, F. (2018). Specificity of interaction between teacher and students in the process of teaching a foreign language. Вопросы науки и образования, (8 (20)), 141-143.
17. Alisherovna, K. S. S. F. N., Amanaliyevich, O. N., & Polatovich, K. S. (2025). MECHANISMS OF IONIZING RADIATION-INDUCED DAMAGE TO CELLS AND DNA. SHOKH LIBRARY, 1(13).

18. Dusaliyev, F. M., & Sh, B. M. (2026). CLINICAL COURSE AND DIAGNOSTIC APPROACHES OF ANORECTAL MALFORMATIONS ASSOCIATED WITH RECTOURETHRAL FISTULAS IN BOYS. Shokh Articles Library, 1(1).
19. Sh, B. M. (2025). HOMILA ICHI MEKONIYALI PERITONITIN TEKSHIRISH VA DAVOLASHNI TAKOMILASHTIRISH (ADABIYOTLAR SHARHI). Central Asian Journal of Academic Research, 3(11-2), 142-148.
20. Бобоев, М. Ш., & Хайдаров, Н. С. (2025). СИНДРОМ ОБЪЁМНОГО ОБРАЗОВАНИЯ БРЮШНОЙ ПОЛОСТИ У ДЕТЕЙ. Eurasian Journal of Medical and Natural Sciences, 5(10-2), 174-181.
21. Khaidarov, N. S., Sh, B. M., & Dusaliyev, F. M. (2026). POSTOPERATIVE ABDOMINAL ADHESIVE DISEASE IN CHILDREN: CLINICAL EXPERIENCE. Shokh Articles Library, 1(1).
22. Саттаров, Ж. Б., & Бобоев, М. Ш. (2025). ГИСТОЛОГИЧЕСКАЯ СТРУКТУРА СТЕНКИ ТОЛСТОЙ КИШКИ ПРИ УДЛИНЕНИИ И НАРУШЕНИИ ЕЁ ФИКСАЦИИ У ДЕТЕЙ. Eurasian Journal of Medical and Natural Sciences, 5(10-2), 84-92.
23. Бобоев, М. Ш., & Саттаров, Ж. Б. (2025). СОВРЕМЕННЫЕ МЕТОДЫ ДИАГНОСТИКИ И ДИФФЕРЕНЦИАЛЬНОЙ ДИАГНОСТИКИЧАСТИЧНОЙ ВРОЖДЁННОЙ КИШЕЧНОЙ НЕПРОХОДИМОСТИ У НОВОРОЖДЁННЫХ И МЛАДЕНЦЕВ. Eurasian Journal of Medical and Natural Sciences, 5(10-2), 76-83.
24. Бобоев, М. Ш., & Саттаров, Ж. Б. (2025). СОВРЕМЕННЫЕ МЕТОДЫ ДИАГНОСТИКИ И ДИФФЕРЕНЦИАЛЬНОЙ ДИАГНОСТИКИЧАСТИЧНОЙ ВРОЖДЁННОЙ КИШЕЧНОЙ

НЕПРОХОДИМОСТИ У НОВОРОЖДЁННЫХ И МЛАДЕНЦЕВ. Eurasian Journal of Medical and Natural Sciences, 5(10-2), 76-83.

25. Sh, B. M. (2025). YANGI TUG ‘ILGAN CHAQALOQLAR VA GO ‘DAKLARDA UCHRAYDIGAN QISMAN TUG ‘MA ICHAK TUTILISHINI ZAMONAVIY DIAGNOSTIK TAKTIKASINI TANLASH. Central Asian Journal of Academic Research, 3(11-2), 136-141.
26. Саттаров, Ж. Б., & Бобоев, М. Ш. (2025). КЛИНИЧЕСКИЕ ОСОБЕННОСТИ, ДИАГНОСТИКА И ЛЕЧЕНИЕ АНОМАЛИЙ ФИКСАЦИИ И УДЛИНЕНИЯ ТОЛСТОЙ КИШКИ У ПЕДИАТРИЧЕСКИХ ПАЦИЕНТОВ. Eurasian Journal of Medical and Natural Sciences, 5(10-2), 93-101.
27. Sh, B. M. (2025). Intrauterine meconium peritonitis (literature review). Eurasian Journal of Medical and Natural Sciences, 5(10-2), 46-51.
28. Sh, B. M. (2025). Cystic duplication of the stomach in children. Web of Medicine: Journal of Medicine. Practice and Nursing, 3(1), 367-371.
29. Турсунова, О. А., & Шарапов, Б. У. (2017). ИЗУЧЕНИЕ ЧАСТОТЫ ЗАБОЛЕВАЕМОСТИ ГЕМОРРАГИЧЕСКИМ ВАСКУЛИТОМ У ДЕТЕЙ. In INTERNATIONAL INNOVATION RESEARCH (pp. 236-239).
30. Шарипова, З. У., Ашурова, Д. Т., & Турсунова, О. А. (2017). Эффективность ступенчатой антибактериальной терапии в лечении пневмонии у детей. Молодой ученый, (16), 102-104.
31. Ашурова, Д. Т., & Садирходжаева, А. А. (2018). Особенности клинической симптоматики поражения сердечно-сосудистой системы при СД 1 типа у детей. Проблемы науки, (2 (26)), 69-73.

32. Садирходжаева, А. А., & Ашурова, Д. Т. (2019). Особенности ранней диагностики диабетической кардиомиопатии во взаимосвязи с кардиологическими маркёрами у детей с сахарным диабетом 1. Уральский медицинский журнал, (8), 22-24.
33. Садирходжаева, А. А., Ашурова, Д. Т., & Шарапов, Б. У. (2019). ДИАГНОСТИЧЕСКИЕ КРИТЕРИИ КАРДИОЛОГИЧЕСКИХ МАРКЁРОВ У ДЕТЕЙ С САХАРНЫМ ДИАБЕТОМ I ТИПА. Новый день в медицине, (2), 50-52.
34. Садирходжаева, А. А., & Ашурова, Д. Т. (2019). Особенности состояния кардиологических маркёров в ранней диагностики диабетической кардиомиопатии у детей с сахарным диабетом 1 типа. Austrian Journal of Technical and Natural Sciences, (3-4), 3-7.
35. Садирходжаева, А. А., & Ашурова, Д. Т. (2022). hs-CRP в сыворотке крови как маркер асептического воспаление стенок сосудов у детей с сахарным диабетом 1 типа. In Молодые ученые-медицине (pp. 109-113).
36. Ахмедова, Д. И., Ишниязова, Н. Д., Салихова, Г. У., & Ашурова, Д. Т. (2012). Особенности психологического развития детей дошкольного возраста. Педиатрия. Илмий-амалий журнал, 38.
37. Ахмедова, Д. И., & Ашурова, Д. Т. (2012). Влияние интегрированного подхода по профилактике микронутриентной недостаточности на некоторые показатели физического развития детей в возрасте 3 лет Республики Каракалпакстан. Педиатрия. Илмий-амалий журнал, 34.
38. Садирходжаева, А. А., Турсунова, О. А., & Шарипова, З. У. (2018). Влияние кислородтранспортной системы крови на тканевую гипоксию у детей с сахарным диабетом I типа. Молодой ученый, (8), 48-51.

39. Robson, M., Im, S. A., Senkus, E., et al. (2017). Olaparib for metastatic breast cancer in patients with a germline BRCA mutation. New England Journal of Medicine, 377, 523–533.
40. Daly, M. B., Pal, T., Berry, M. P., et al. (2020). Genetic/familial high-risk assessment: Breast, ovarian, and pancreatic cancer. Journal of the National Comprehensive Cancer Network, 18(4), 380–391.
41. National Comprehensive Cancer Network (NCCN). (2023). Clinical practice guidelines in oncology: Genetic/familial high-risk assessment. NCCN Guidelines.
42. Kuchenbaecker, K. B., McGuffog, L., Barrowdale, D., et al. (2016). Evaluation of polygenic risk scores for breast cancer in BRCA mutation carriers. Journal of the National Cancer Institute, 109(7).
43. Rebbeck, T. R., Friebel, T., Lynch, H. T., et al. (2004). Bilateral prophylactic mastectomy reduces breast cancer risk in BRCA mutation carriers. Journal of Clinical Oncology, 22(6), 1055–1062.
44. Domchek, S. M., Friebel, T. M., Singer, C. F., et al. (2010). Association of risk-reducing surgery in BRCA mutation carriers with cancer risk and mortality. JAMA, 304(9), 967–975.