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REFERRAL CRITERIA AND CLINICAL APPLICATION OF PREVENTIVE BRCA1/BRCA2 GENETIC TESTING IN BREAST AND OVARIAN CANCER: A NARRATIVE REVIEW

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ABSTRACT

Background: Patients inheriting BRCA1 or BRCA2 mutations face significantly elevated risks of breast, ovarian, and pancreatic cancers, and melanoma. Identifying high-risk individuals enables timely prevention and selection of targeted therapies.
Objectives: This review summarizes genetic testing referral guidelines, discusses BRCA1/2 roles in DNA repair, presents modern diagnostic methods, and addresses preventive strategies for mutation carriers.
Methods/Scope: A comprehensive examination of clinical practice guidelines (including NCCN criteria), diagnostic paradigms (NGS, MLPA, Sanger sequencing), risk assessment models, targeted therapies, surgical risk-reduction strategies, and lifestyle factors was conducted.
Results/Discussion: Genetic testing enables early carrier identification for prevention and qualifies cancer patients for targeted PARP inhibitor therapy. Next-Generation Sequencing (NGS) has replaced Real-time PCR—which only detected common founder mutations—achieving diagnostic sensitivity above 99%. Qualification relies on pedigree analysis and risk evaluation. Prophylactic bilateral mastectomy and salpingo-oophorectomy reduce breast and gynecological cancer risks by 90–95%, while intensified surveillance with MRI ensures early detection. Young BRCA1/2 carriers face challenges regarding fertility preservation, surgical menopause, altered body image, and psychological stress, requiring multi-specialty care. Male carriers also require structured prostate and breast monitoring.
Conclusion: Early identification of BRCA1/2 carriers by primary care physicians, gynecologists, and oncologists is essential for optimizing risk reduction, surveillance, and personalized oncology. Population-wide screening in individuals with unburdened family histories remains unindicated.

KEYWORDS

BRCA1, BRCA2, Hereditary Breast And Ovarian Cancer (HBOC), Next-Generation Sequencing (NGS), PARP Inhibitors, Prophylactic Salpingo-Oophorectomy, Risk-Reducing Mastectomy, Cascade Testing

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Introduction

Breast cancer is the most frequently diagnosed malignant neoplasm among women worldwide. Despite its high prevalence, the underlying cause in the majority of individual cases remains unknown. Among the most clinically recognized risk factors are advanced age, carriage of pathogenic mutations in specific genes - predominantly BRCA1 and BRCA2 - as well as family history of breast cancer, particularly when diagnosed at a young age [1, 2]. Additionally, well-documented risk factors include early age at menarche, late age at menopause, advanced age at first full-term live birth, prolonged use of hormone replacement therapy (HRT), overweightness, obesity, exposure to ionizing radiation (specifically thoracic radiotherapy administered before 30 years of age), and certain benign proliferative breast lesions [2, 3].

The BRCA1 and BRCA2 genes produce proteins responsible for repairing damaged DNA [1, 15]. Every individual possesses two copies of each gene, one inherited from each parent [1]. Inheriting a mutated or non-functional copy of either gene significantly increases the likelihood of developing specific malignancies, most commonly breast cancer and ovarian cancer [1, 9]. Cumulatively, over 60% of women who inherit a damaged BRCA1 or BRCA2 gene will develop breast cancer during their lifetime [1, 9]. Furthermore, approximately 39% to 58% of women with a BRCA1 mutation and roughly 13% to 29% of women with a BRCA2 mutation will develop ovarian cancer [1, 9]. Beyond breast and gynecological malignancies, inheriting a damaged copy of BRCA1/2 is associated with an elevated risk of pancreatic cancer, prostate cancer, and cutaneous melanoma [1, 11]. Experts recommend considering BRCA1 and BRCA2 genetic testing for individuals at higher risk of carrying a harmful mutation than the general population [2, 9]. For individuals with a significant family history, genetic testing using multi-gene panels should be offered systematically [2, 5].

Objectives and Functional Roles of BRCA Testing

Clinical Utility of BRCA Status Evaluation

In modern medicine, determining BRCA gene status serves a dual purpose [2, 7]:

Firstly, it serves a preventive function by identifying high-risk patients with a family history of breast cancer [2, 9]. Early knowledge of mutation carriage enables the implementation of intensified screening protocols such as initiating magnetic resonance imaging (MRI) surveillance at age 25 or the consideration of risk-reducing surgical procedures [2, 3].

Secondly, it plays a key role in guiding treatment selection [7, 16]. Determining BRCA gene status identifies eligible patients for targeted treatment with PARP inhibitors [16, 17, 18]. Furthermore, carrying a BRCA mutation directly influences surgical management decisions regarding the appropriate extent of oncological resection [2, 7].

Functional Rationale of BRCA1 and BRCA2 Genes

The BRCA1 and BRCA2 genes are functionally interconnected because they both play a fundamental role in maintaining genomic stability through the homologous recombination repair pathway [15]. However, they perform distinct tasks when responding to DNA damage [15]. BRCA1 functions as a master, pleiotropic coordinator; its primary responsibilities include recognizing double-strand DNA breaks, activating cell cycle checkpoints, and recruiting downstream repair machinery [15]. Conversely, BRCA2 acts as a direct molecular mediator of homologous recombination [15]. It binds to the RAD51 recombinase and coordinates the assembly of nucleoprotein filaments on single-stranded DNA, thereby enabling accurate and error-free DNA repair [15].

Organizational Framework and Diagnostic Workflow

Clinical Referral Pathways

Certified clinical genetics specialists should provide clinical genetics counseling and diagnostic management in genetics clinics [4, 19]. In Poland, any licensed physician practicing under an agreement with the National Health Fund (NFZ) can legally refer a patient to a clinical genetics clinic [4, 19]. In daily practice, primary care physicians most frequently initiate referrals based on a detailed family history, such as recurring cases of ovarian, breast, or pancreatic cancer within a patient's family [4, 19]. A gynecologist may similarly issue a referral based on family history or concerning physical findings in the reproductive organs [4, 19]. Oncologists refer patients with an established cancer diagnosis, such as women diagnosed with breast cancer before age 50 [4, 7]. The oncologist orders genetic testing both to assess prognosis and preventive options, as well as to select targeted therapies, including PARP inhibitors [7, 18].

Although any physician can issue an initial referral to a clinical genetics clinic, the final decision regarding genetic testing and its scope is made by a certified specialist in clinical genetics or a physician undergoing specialty training in clinical genetics [4, 19]. The clinical geneticist analyzes the patient's pedigree and uses risk calculation tools alongside clinical guidelines to evaluate indications for testing, and qualifies the patient for the appropriate test [4, 11]. During the consultation, the objective of the examination is thoroughly explained, and the patient must provide formal written informed consent before testing [4, 19].

Laboratory Procedure and Sample Processing

The diagnostic procedure typically involves drawing a peripheral venous blood sample from the cubital vein into a tube containing EDTA anticoagulant to isolate genomic DNA [7]. The sample is subsequently analyzed in a genetic laboratory where DNA is extracted, and Next-Generation Sequencing (NGS) of the BRCA1 and BRCA2 genes is performed [7]. When clinically indicated, testing is supplemented with Multiplex Ligation-dependent Probe Amplification (MLPA) [7]. Finally, the results are thoroughly discussed during a consultation with a clinical geneticist [4, 19]. If a mutation is identified, a personalized preventive screening program is formulated, surgical options for risk reduction are reviewed, and the necessity of cascade testing for family members is carefully evaluated [2, 10].

Criteria for Referral to BRCA1/BRCA2 Genetic Testing

Family History and Risk Assessment Models

Evaluating family history is the most critical factor in qualifying a patient for genetic testing [2, 11]. However, in approximately 10% of breast cancer patients, a complete family history cannot be obtained due to a lack of information regarding relatives or limited family size [9, 11]. In most countries, a calculated risk threshold of 10% or greater - determined using computer-based risk estimation software such as Gail, Claus, Cuzick-Tyrer, BRCAPRO, BOADICEA, Myriad 1 and 2, or Manchester models - is considered sufficient to qualify for genetic testing [2, 11]. Key personal and family history features associated with an increased likelihood of harboring a BRCA mutation include breast cancer diagnosis before the age of 40, multiple cases of breast or ovarian cancer within the family, and cases of multiple primary tumors in the same individual, particularly when one is ovarian cancer [2, 11]. Additional qualification criteria include a family history of bilateral breast cancer, male breast cancer, premenopausal triple-negative breast cancer, medullary or atypical medullary carcinomas, Ashkenazi Jewish ancestry, or a previously documented BRCA mutation within the family [5, 11].

Summary of NCCN Eligibility Criteria for BRCA1/BRCA2 Testing

Based on National Comprehensive Cancer Network (NCCN) guidelines [5, 6, 11, 14], eligibility criteria are summarized as follows:

Table 1. Summary of NCCN Eligibility Criteria for BRCA1/BRCA2 Testing

Malignancy / Context	Qualification Criteria for Genetic Testing
Ovarian Cancer	Every patient diagnosed with non-mucinous epithelial ovarian, fallopian tube, or primary peritoneal cancer, regardless of age or family history (germline and/or somatic testing offered at diagnosis) [5,11].
Female Breast Cancer	• Diagnosis at or before age 50 [5,6] • Triple-negative breast cancer (TNBC) diagnosed at or before age 60 [5,6] • Bilateral or multifocal breast cancer [5,11]. • Medullary or atypical medullary carcinoma [5,6] • Testing required to guide targeted therapy (e.g., PARP inhibitors) [6,18].
Male Breast Cancer	Every male patient diagnosed with breast cancer, regardless of age [6,14].
Other Malignancies	• Pancreatic adenocarcinoma in any patient [11,14] • High-grade or metastatic prostate cancer [6,14].
Ancestry & Pedigree	• Ashkenazi Jewish ancestry with any personal or family history of breast or ovarian cancer [5,11]. • Documented BRCA1/2 mutation in a 1st- or 2nd-degree relative [10,11]. • Clustering of cancers in the family (at least 2 cases of breast, ovarian, or pancreatic cancer on the same side of the family) [5,11].

Inheritance Patterns and Cascade Diagnostics

Mode of Inheritance

Hereditary Breast and Ovarian Cancer (HBOC) syndrome, associated with mutations in the BRCA1 and BRCA2 genes, is inherited in an autosomal dominant manner [1, 9]. Offspring of an individual with a pathogenic germline variant in BRCA1 or BRCA2 have a 50% chance of inheriting it [1, 9]. The subsequent risk of developing cancer in offspring who inherit the pathogenic variant depends on multiple factors, including variant penetrance, biological sex, and current age [1, 2]. Mutations can be transmitted through both maternal and paternal lines [1, 10]. Men act as obligate carriers who can pass the mutation to their children, while facing elevated personal risks for male breast cancer, prostate cancer, and pancreatic cancer [6, 14].

Cascade Genetic Testing in Affected Families

Cascade testing is a sequential screening process offered to family members who may share the same genetic condition based on pedigree relationships and inheritance patterns [10]. The primary goal of cascade testing is to identify asymptomatic carriers as early as possible before cancer develops [10]. This allows individuals to be enrolled in intensive surveillance programs, such as regular breast MRI screening before the age of 30 for female BRCA1/2 mutation carriers [2, 10].

The cascade procedure begins when a specific pathogenic variant - such as a BRCA1 or BRCA2 mutation - is identified in a family member [10]. Once the exact mutation is known, close relatives, starting with first-degree relatives (parents, children, siblings), are tested specifically for that single variant [10]. If mutation carriage is confirmed, intensive preventive surveillance, early diagnostics, and risk-reducing measures are promptly initiated [2, 10]. Unlike the index case (proband) - frequently an individual already affected by breast or ovarian cancer who requires full-gene NGS sequencing - relatives undergo targeted testing for the single known familial mutation, making the process rapid and cost-effective [7, 10].

Diagnostic Pathway Evolution: Real-Time PCR to NGS

Historically in Poland, genetic testing for BRCA1/2 mutations relied on diagnostic algorithms using targeted Real-time PCR techniques [7, 8]. However, this approach had clear limitations, as it detected only common founder mutations - typically 3 to 5 population-specific variants [7, 20]. Although cost-effective, this method identified only about 50% of pathogenic variants in BRCA1 and omitted analysis of the BRCA2 gene entirely [7, 20].

Currently, national and international guidelines strongly recommend Next-Generation Sequencing (NGS) as the primary diagnostic method [5, 7]. Unlike real-time PCR, NGS enables comprehensive sequencing of all coding regions of both BRCA1 and BRCA2, achieving diagnostic sensitivity exceeding 99% [7]. This comprehensive profiling eliminates the risk of false-negative results associated with founder mutation panels [7]. NGS is an indispensable tool for treatment optimization by facilitating timely qualification for targeted PARP inhibitor therapy [7, 18]. Targeted Sanger sequencing and MLPA techniques remain widely utilized in cascade testing for family members and for detecting large genomic rearrangements [7, 10]. Rapid acquisition of a complete BRCA1/2 mutation profile is critical to ensure patients receive prompt, optimal targeted care [7, 18].

Using Next-Generation Sequencing (NGS) instead of Real-time PCR in routine clinical diagnostics has significantly improved the detection rate of BRCA1 and BRCA2 mutations; however, it presents new economic and organizational challenges [7]. Although the cost of performing NGS testing has decreased substantially over the past 10 years, it remains a method requiring specialized equipment, dedicated software, and highly qualified personnel [7]. The waiting time to obtain test results represents another major issue, currently ranging from several weeks to a few months [7, 8]. When genetic testing is performed in a patient already diagnosed with cancer, delays in acquiring results can directly impact therapeutic decisions, such as delaying the initiation of PARP inhibitor therapy or postponing the planning of the appropriate surgical extent [7, 18]. Establishing fast-track diagnostic pathways for oncology patients whose genetic test results directly influence therapy selection is of paramount importance [4, 7]. Furthermore, healthcare systems strive to optimize reimbursement criteria to ensure equitable access to modern molecular diagnostics while maintaining control over public expenditures [4, 7].

Therapeutic Implications of BRCA Mutation Status

Antineoplastic agents that induce DNA damage, such as cisplatin, demonstrate notable efficacy in treating tumors in individuals carrying a BRCA1 or BRCA2 mutation [16]. This heightened sensitivity occurs because mutated BRCA genes impair homologous recombination DNA repair mechanisms [15, 16]. Cancer cells unable to resolve DNA lesions induced by cisplatin or similar platinum-based chemotherapies are rendered significantly more vulnerable to cell death [16].

PARP inhibitors are highly effective in treating tumor cells harboring deleterious BRCA1 or BRCA2 alterations, regardless of whether these mutations are inherited or somatically acquired [16, 17]. The mechanism of action of PARP inhibitors involves blocking single-strand DNA break repair, leading to double-strand breaks during replication that BRCA-deficient cells cannot resolve [16]. Currently, four PARP inhibitors approved by the U.S. Food and Drug Administration (FDA) - olaparib, rucaparib, niraparib, and talazoparib - are utilized to treat specific malignancies harboring BRCA1/2 alterations [16, 17, 18].

Individuals who inherit a BRCA1 or BRCA2 mutation display a higher incidence of triple-negative breast cancer (TNBC) compared to non-carriers or the general population [5, 18]. TNBC lacks estrogen receptors, progesterone receptors, and HER2 overexpression [18]. Due to the absence of these therapeutic targets, triple-negative tumors are more challenging to treat and carry a worse prognosis [18]. When genetic testing identifies a BRCA1/2 mutation, patients should consult their physician to determine whether the variant is hereditary, which is essential for estimating cancer risks in close biological relatives [9, 10].

Preventive Management and Risk-Reduction Strategies

Individuals in high-risk categories, including BRCA1/2 mutation carriers, should evaluate surgical and non-surgical preventive options [2, 3]. Prospective and retrospective analyses demonstrate that bilateral prophylactic mastectomy in BRCA1/2 carriers reduces breast cancer risk by 90% to 95% [2, 3, 19]. Similarly, prophylactic removal of the ovaries and fallopian tubes significantly lowers ovarian cancer risk and reduces all-cause mortality before age 70 [2, 3, 19]. Following completion of childbearing and reaching 35 to 40 years of age, prophylactic bilateral adnexectomy is recommended [2, 3, 19].

Surgical risk reduction represents the most effective form of prevention in BRCA1/2 carriers [2, 3]. Bilateral prophylactic mastectomy involves removing glandular tissue from both breasts, frequently accompanied by immediate reconstruction preserving the nipple-areolar complex [2, 19]. The minor residual risk of 5% to 10% stems from microscopic residual glandular cells [2]. Prophylactic salpingo-oophorectomy involves removing both ovaries and fallopian tubes, yielding an approximate 90% reduction in ovarian and fallopian tube cancer risk [2, 3]. Guidelines recommend this procedure between ages 35 and 40 for BRCA1 carriers upon completing reproductive plans, whereas BRCA2 carriers may defer surgery until ages 40 to 45 due to the later average onset of malignancy [2, 3, 11]. Current research indicates that a substantial proportion of high-grade serous epithelial ovarian cancers originate in the fimbriae of the fallopian tubes, fully justifying complete adnexal resection [2, 3].

Due to the elevated risk of contralateral primary breast cancer, BRCA1/2 carriers have clear indications for specialized oncological surveillance utilizing breast MRI [2, 8]. Its sensitivity and specificity in detecting early-stage breast lesions far exceed those of mammography or ultrasonography [2, 8]. Mutation carriers also face a higher probability of multifocal disease, which often remains occult on mammograms [2, 8].

Intensified screening for women in the high-risk group achieves a combined sensitivity of approximately 95% [2, 19]. Women should begin monthly breast self-examinations at age 18 after proper instruction [19]. Starting at the age of 25, clinical breast examinations should be performed by a healthcare professional every 6 to 12 months [2, 19]. Because mammography before age 30 nearly doubles radiation-induced cancer risks in BRCA carriers, breast MRI is the preferred screening modality in this age group [2, 5]. Between ages 25 and 30, MRI is performed annually (alternating with mammography when indicated), supplemented by annual breast ultrasonography [2, 19]. Dense breast tissue in young women further reduces mammographic sensitivity [2]. From age 35, screening includes transvaginal ultrasonography and serum CA-125 biomarker monitoring every 6 months, alongside annual transvaginal gynecological examinations [2, 19]. However, gynecological screening does not substitute for prophylactic surgery [2, 3].

Lifestyle and dietary modifications also play an important role [13]. American Cancer Society guidelines emphasize regular physical exercise, weight management, minimizing alcohol consumption, encouraging lactation, and avoiding hormone replacement therapy [13]. Cigarette smoking acts as a systemic mutagen that further impairs DNA repair capacity; prolonged smoking in BRCA1/2 carriers increases the risk of breast, pancreatic, and gynecological cancers [13]. Adult weight gain and obesity elevate postmenopausal breast cancer risk, whereas moderate-to-vigorous physical activity reduces breast cancer risk by 25% across pre- and postmenopausal states [13]. A diet rich in vegetables, fruits, poultry, fish, and low-fat dairy may selectively lower the risk of hormone receptor-negative breast cancers [13]. Additionally, breastfeeding for at least one year exerts a strong protective effect in BRCA1 carriers, significantly lowering breast cancer risk [13].

Psychosocial Considerations and Male Mutation Carriers

Challenges in Young Female Carriers

Recommending prophylactic interventions such as bilateral mastectomy or salpingo-oophorectomy creates complex life dilemmas for young carriers, particularly regarding family planning [2, 12]. Ovarian and fallopian tube excision induces immediate surgical menopause and irreversible fertility loss, necessitating thorough discussions regarding fertility preservation options prior to surgery [2, 12].

Carrying a BRCA1/2 mutation imposes a major psychological burden [2, 9]. Carriers often experience chronic health-related anxiety that heightens before routine screening appointments [2, 9]. Specialized psycho-oncological support within clinical genetics units should be systematically provided throughout the diagnostic process and follow-ups [2, 4]. Furthermore, bilateral mastectomy significantly alters body image and self-esteem; modern immediate breast reconstruction options help preserve psychological well-being and quality of life [2, 19].

BRCA Mutations in Male Carriers

Although BRCA1 and BRCA2 mutations are primarily associated with female breast and ovarian cancers, autosomal dominant inheritance means men have an equal 50% probability of inheriting and transmitting pathogenic variants [1, 10]. Men carrying a damaged BRCA2 gene face elevated risks of male breast cancer, pancreatic cancer, and early-onset prostate cancer [6, 14]. According to NCCN guidelines, male carriers should undergo prostate-specific antigen (PSA) testing and urological examinations starting at age 40, along with annual clinical breast examinations starting at age 35 [6, 14].

Conclusions

Early identification of BRCA1/2 mutation carriers is essential for effective cancer prevention, structured active surveillance, and individualized treatment [2, 7]. Primary care physicians, gynecologists, and oncologists must proactively identify patients who meet the risk criteria and refer them to clinical genetics services [4, 19]. This structured pathway ensures optimal risk reduction, rigorous active surveillance, and timely access to targeted therapies - such as PARP inhibitors - should malignancy occur [7, 18]. Specific management guidelines created for BRCA1/2 carriers, including early-onset clinical breast examinations and specialized imaging (MRI, ultrasound, mammography), must be followed strictly [2, 5]. Population-wide genetic testing in healthy women without a suggestive family history remains unindicated under current guidelines [2, 9].

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