POSITION STATEMENT
Hereditary Breast and Ovarian Cancer Risk Assessment

The National Association of Nurse Practitioners in Women’s Health (NPWH) supports the role of women’s health nurse practitioners (WHNPs) in providing hereditary breast and ovarian cancer (HBOC) risk assessment. At a minimum, HBOC assessment should include the patient’s personal cancer history; cancer histories of maternal and paternal first- and second-degree relatives, with descriptions of the types of primary cancers and ages at onset; any Eastern or Central European Jewish ancestry; and the results of cancer predisposition testing in any relative. Because family history is dynamic, the assessment should be reviewed and updated regularly. The goal of HBOC risk assessment is to identify those who may benefit from genetic counseling, genetic testing, enhanced surveillance, or other risk-management strategies.

WHNPs should be knowledgeable about indicators of an increased risk for HBOC, as put forth by the National Comprehensive Cancer Network (NCCN). Individuals assessed as being at increased risk should have access to genetic counseling by clinicians with training and expertise in cancer genetics. These specialists can provide genetic testing if indicated and desired, psychosocial support, and evidence-based management that depends on identified risk and genetic testing results if such testing is done. Resources for locating cancer genetics specialists include the National Society of Genetic Counselors website and the National Cancer Institute’s NCI Cancer Genetics Services Directory.

A system should be established within the practice setting for referral, consultation, and/or collaboration to ensure that patients have timely access to genetic counseling services and subspecialty follow-up. It is recognized that with an increase in HBOC risk assessment in primary care settings, there can be barriers to access to the limited number of genetics specialists, especially in rural areas. Such barriers may be addressed through avenues such as telegenetic counseling and collaborative models that support WHNPs and other primary care providers in providing in-depth risk assessment, pre-test counseling, and genetic testing. Therefore, NPWH opposes reimbursement requirements mandating that pre-test counseling be provided only by an individual certified in genetic counseling before genetic testing can be ordered. Such requirements unduly limit access to timely care for some individuals.

Primary care providers with appropriate training and skills, including WHNPs, may provide pre-test counseling and order HBOC genetic testing. Obtaining such knowledge and skills, as well as keeping up to date on evolving cancer genetics knowledge and testing technology, requires additional training beyond that received in a WHNP program. An evidence-based protocol established according to guidelines provided by nationally recognized organizations such as NCCN must be followed to ensure that all recommended components of assessment, counseling, informed consent, appropriate testing, and follow-up are provided. State and federal regulations and laws for informed consent with regard to genetic testing and reimbursement also must be followed. WHNPs can help ensure that patients receive timely, evidence-based care when identified as being at risk for HBOC. The WHNP should consult with or refer the individual to a specialist in cancer genetics when the...
patient’s history or test results present a complex situation or when the patient requests such a referral.

NPWH provides leadership and collaborates with other organizations and agencies to deliver education that prepares WHNPs with the knowledge and skills to provide HBOC risk assessment, individualized counseling, and genetic testing in collaboration with specialists in cancer genetics. In addition, NPWH provides leadership in monitoring and supporting reimbursement and other policies that ensure qualified WHNPs are able to counsel patients on HBOC risks and to order and interpret genetic tests. Furthermore, NPWH collaborates with other organizations and agencies to support research to better inform healthcare providers on best practice for identifying individuals at risk for HBOC and for counseling, testing, and risk management.

BACKGROUND

The American Cancer Society (ACS) projects that in 2022, a total of 287,850 new cases of breast cancer will be diagnosed in women in the United States and 43,250 women will die of breast cancer. Breast cancer 2022 projections for men are for a total of 2,710 new cases and 503 deaths. The ACS also projects that in 2022, a total of 19,800 new cases of ovarian cancer will be diagnosed and 12,810 deaths will be caused by ovarian cancer.\(^\text{10}\)

The majority of breast and ovarian cancers are not related to unique identifiable risk factors, although certain individual, familial, reproductive, and lifestyle factors have been associated with increased risk.\(^\text{11}\) Approximately 5% to 10% of breast cancers and 20% to 25% of ovarian cancers are associated with a hereditary predisposition from an inherited pathogenic variant (previously called a mutation).\(^\text{6}\)

Over the years, HBOC has largely been explained by pathogenic variants occurring in \textit{BRCA1} or \textit{BRCA2} genes and has been described as HBOC syndrome.\(^\text{12}\) Because of the autosomal dominant inheritance pattern of this condition, a person has a 50% chance of passing a pathogenic variant in \textit{BRCA1} or \textit{BRCA2} to offspring, regardless of gender. Approximately 1 in 300 to 500 persons in the general population carry pathogenic variants in \textit{BRCA1} or \textit{BRCA2}.\(^\text{6}\)

The lifetime cumulative risk (to age 70) for breast cancer is estimated to be 45% to 65% for women with a pathogenic variant in \textit{BRCA1} or \textit{BRCA2}, as compared with a 13% risk for unaffected women.\(^\text{13–16}\) The lifetime risk for ovarian cancer is estimated to be 39% to 65% for women with a pathogenic variant in \textit{BRCA1} and 10% to 17% for women with a pathogenic variant in \textit{BRCA2}, as compared with a 1% to 2% risk for unaffected women.\(^\text{13–16}\) In many cases, the pathogenic variants found in \textit{BRCA1} and \textit{BRCA2} result in cancer onset at an earlier age than would be expected with cancers not associated with hereditary cancer genes. Although pathogenic variants in \textit{BRCA1} and \textit{BRCA2} are responsible for the majority of HBOC cases, a growing number of other genes have been associated with an increased risk for breast cancer and/or ovarian cancer.\(^\text{4,5,17–19}\) NCCN provides information on more than two dozen known hereditary cancer genes (eg, \textit{PALB2}, \textit{CHEK2}, \textit{CDH1}) that increase the risk for breast cancer and/or ovarian cancer.\(^\text{6}\)

Risk assessment that includes a patient’s personal and family history of cancer is the preliminary step in determining if that person might benefit from genetic counseling and genetic testing.\(^\text{1,2,4–20}\) Several screening tools/risk prediction models designed to identify a personal and/or family history that may be associated with inherited cancer susceptibility are available.\(^\text{1,6,7}\) Each tool/model has its strengths and limitations that must be considered in the context of the individual patient.\(^\text{7}\)

Strategies for conducting preliminary HBOC risk assessment include questionnaires completed prior to or during the visit.\(^\text{5,20}\) NCCN describes the criteria that warrant further risk assessment, genetic counseling, and consideration for genetic testing.\(^\text{1}\)

Genetic counseling provides an opportunity to further assess risk and explore whether a patient is a candidate for or desires genetic testing for cancer susceptibility. Counseling is valuable for individuals with increased breast and ovarian cancer risk even when they are not a candidate for or do not desire genetic testing, because they may benefit from enhanced cancer surveillance and other risk-management strategies.\(^\text{2,5,7}\) The comprehensive genetic counseling process should include a review of personal and family history or pedigree; a detailed risk assessment; family history or pedigree; a detailed risk assessment; genetic testing, and consideration for genetic testing.\(^\text{1}\)

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When genetic testing is indicated, choices include single-gene testing and multi-gene panel testing. Compared with single-gene testing, multi-gene panel testing with next-generation sequencing technology increases the rate of detection of pathogenic variants and is a more time- and cost-effective approach. NCCN provides management guidelines for the care of patients found to have certain pathogenic variants that can be identified by multi-gene panel testing. However, standard management guidelines are not yet available for all genes. In addition, multi-gene panel testing can result in a higher likelihood of detecting one or more variants of uncertain clinical significance (VUS). Patients need to be informed of the benefits and limitations of multi-gene panel testing and the meaning of VUS before testing is performed.

Individualized cancer risk-reducing strategies and enhanced surveillance schedules should be established for patients who test positive for pathogenic variants that place them at risk for HBOC. Risk-reducing strategies and enhanced surveillance schedules also are indicated for patients with a personal or family history concerning for HBOC syndrome, even when no pathogenic variant is detected or only a VUS is identified. Implications for family members regarding testing and risk modification should be addressed if a pathogenic variant is identified.

In some situations, risk-reducing strategies and enhanced surveillance for transgender and nonbinary individuals who have pathogenic variants in BRCA1 and BRCA2 genes follow the same NCCN guidelines as for cisgender individuals. However, there is insufficient evidence to know whether gender-affirming hormone therapy alters breast cancer risk and how this affects decisions about risk-reducing strategies and enhanced surveillance. More research is needed to establish evidence-based guidelines inclusive of transgender individuals.

**IMPLICATIONS FOR WOMEN’S AND GENDER-RELATED HEALTHCARE AND WHNP PRACTICE**

Identifying individuals who may benefit from HBOC genetic counseling, genetic testing, enhanced surveillance, and other cancer risk-management strategies is essential to improve health outcomes. WHNPs are ideally positioned to conduct HBOC risk assessment during wellness and other visits. HBOC risk assessment can be completed as part of the routine health history or using HBOC risk-assessment questionnaires completed by the patient during or prior to the visit. WHNPs with appropriate knowledge and skills regarding HBOC risk assessment and genetic testing indications, implications, and limitations can provide counseling, order and interpret genetic tests if indicated and desired by the patient, and discuss individualized enhanced surveillance recommendations and risk-management strategies. WHNPs who have appropriate training can fill an unmet need to increase availability and accessibility to timely counseling and testing of individuals who are identified as being at risk for HBOC.

**RECOMMENDATIONS**

WHNPs should:

- Conduct preliminary HBOC risk assessment with all adult individuals for whom they provide healthcare and update the assessment regularly.
- Know the indicators for an increased risk for HBOC in a preliminary risk assessment.
- Establish resources for referral, consultation, and/or collaboration when an increased risk for HBOC is identified.
- If planning to provide HBOC counseling and genetic testing, obtain additional training and skills, follow evidence-based guidelines, and adhere to state and federal regulations and laws for informed consent and reimbursement.

NPWH will provide leadership and resources to ensure that:

- Educational programs for WHNP students impart evidence-based knowledge and skill building for development of competencies to conduct preliminary HBOC risk assessment.
- Continuing education programs are available for WHNPs to obtain evidence-based knowledge and competencies to provide HBOC counseling and genetic testing.
- WHNPs have information on available programs for certification in advanced genomics or advanced genetics in nursing.
REFERENCES


Web resources

A. nsgc.org/page/find-a-gc-search

B. cancer.gov/cancertopics/genetics/directory

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