

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 risk assessment should include the woman's personal cancer history; her maternal and paternal first-, second-, and third-degree relative cancer histories, with descriptions of the types of primary cancers and the ages of onset; any Ashkenazi Jewish ancestry; and the results of any cancer predisposition testing in any relative.¹ This assessment should be reviewed and updated regularly. The goal of HBOC risk assessment is to identify women who may benefit from genetic counseling, genetic testing, enhanced surveillance, or other risk management strategies.¹⁻⁷

WHNPs should be knowledgeable about indicators of an increased risk for HBOC, as put forth by the National Comprehensive Cancer Network (NCCN).⁵ Women 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^A](#) and the [National Cancer Institute's NCI Cancer Genetics Services Directory^B](#).

Primary care providers with appropriate training and skills, including WHNPs, may provide HBOC genetic counseling and testing.^{6,8} Obtaining such knowledge and skills, as well as keeping up to date with 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.^{2,5} State and federal regulations and laws for informed consent with regard to genetic testing and reimbursement must also be followed. WHNPs should con-

sult with or refer women to a specialist in cancer genetics when their history or test results present a complex situation or when they request such a referral.

A system should be established within WHNPs' practice settings for referral, consultation, and/or collaboration to ensure that women have timely access to genetic counseling services and subspecialty follow-up. Access barriers to a specialist in cancer genetics may be addressed through avenues such as telegenetic counseling^{4,9} and collaborative models that support WHNPs in providing in-depth risk assessment, 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 can unduly limit access to timely care.⁸

NPWH will provide leadership and collaborate with other organizations and agencies to deliver education that prepares WHNPs with the knowledge and skills to provide HBOC risk assessment, counseling, and genetic testing in collaboration with specialists in cancer genetics. WHNPs can help ensure that women receive timely, evidence-based care when identified as being at risk for HBOC. In addition, NPWH will provide leadership in monitoring and developing reimbursement and other policies ensuring that qualified WHNPs are able to counsel women regarding HBOC risks and to order and interpret genetic tests. Furthermore, NPWH will collaborate with other organizations and agencies to support research to better inform providers and women 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 2017, a total of 252,710 new cases of breast cancer will be diagnosed in women in the United States and that 40,610 women will die of breast cancer.¹⁰ In addition, the ACS projects that in 2017, a total of 22,440 new cases of ovarian cancer will be diagnosed, with 14,080 deaths caused by ovarian cancer.

Most 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.^{11,12} About 5%-10%

of breast cancers and 20%-25% of ovarian cancers are associated with a predisposition from an inherited pathogenic variant (previously called a mutation).^{2,13,14}

Over the years, HBOC has largely been explained by pathogenic variants occurring in *BRCA1* or *BRCA2* genes, and has been described as HBOC syndrome.¹⁵ Because of the autosomal dominant inheritance pattern of this disease, a person has a 50% chance of passing a pathogenic variant in *BRCA1* or *BRCA2* to offspring, regardless of gender. Approximately 1 in 300-500 persons in the general population and approximately 1 in 40 Ashkenazi Jews carry pathogenic variants in *BRCA1* or *BRCA2*.^{2,6,16}

The lifetime risk (to age 70) for breast cancer is 46%-87% for women with a pathogenic variant in *BRCA1* or *BRCA2*, as compared with a 13% risk for unaffected women. The lifetime risk for ovarian cancer is 39%-63% for women with a pathogenic variant in *BRCA1* and 16.5%-27% for women with a pathogenic variant in *BRCA2*, as compared with a 1%-2% risk for unaffected women.^{2,7,16-19}

Although pathogenic variants in *BRCA1* and *BRCA2* are responsible for most HBOC cases, a growing number of other genes have been found to be associated with an increased risk for breast cancer and/or ovarian cancer. NCCN provides information on more than two dozen known hereditary cancer genes that increase breast and/or ovarian cancer risk.^{5,20} In many cases, the pathogenic variants found in *BRCA1* and *BRCA2* and in the additional hereditary cancer genes result in cancer onset at an earlier age than would be expected with cancers not associated with hereditary cancer genes.^{2,4,7,15}

Risk assessment that includes a woman's personal and family history of cancer is the preliminary step in determining if she might benefit from genetic counseling and genetic testing.^{1,2,4,6,21} Strategies for conducting preliminary HBOC risk assessment include questionnaires sent to patients prior to a visit, questionnaires completed while waiting to see the clinician, and obtaining the information while gathering other elements of the health history.^{6,21}

Several screening tools designed to identify a family history that may be associated with inherited cancer susceptibility are available. Women with positive screening test results should be referred for genetic counseling.^{6,12} NCCN describes the criteria that warrant further risk assessment, genetic counseling, and consideration for genetic testing.⁵

Genetic counseling provides an opportunity to further assess risk and explore whether a woman is a candidate for or desires genetic testing for cancer susceptibility genes. Counseling is valuable for a woman with increased breast and ovarian cancer risk even when she is not a candidate for or does not desire testing, because she may benefit

from enhanced cancer surveillance and other risk-management strategies.^{2,5,12} The comprehensive genetic counseling process should include a review of personal and family history; a detailed risk assessment; psychosocial assessment and support; individualized risk counseling and education; a discussion of genetic testing, including ethical and legal implications; and informed consent.^{5,6,12}

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 women found to have certain pathogenic variants that can be identified by multi-gene panel testing. However, standard management guidelines are not yet available with regard to all of them. In addition, multi-gene panel testing can result in a higher likelihood of detecting one or more variants of uncertain clinical significance (VUS). Women need to be informed of the benefits and limitations of multi-gene panel testing and the meaning of VUS before testing is performed.^{3,13}

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

Implications for women's healthcare and WHNP practice

Identifying women 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 well-woman 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 prior to or during 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 woman, and discuss individualized enhanced surveillance recommendations and risk-management strategies. NPWH recognizes that

WHNPs who have appropriate training can fill an unmet need to increase availability and accessibility to timely counseling and testing of women who are identified as being at risk for HBOC.

Recommendations

WHNPs should:

- Conduct preliminary HBOC risk assessment with all women for whom they provide healthcare and update the assessment regularly.
- In a preliminary risk assessment, know the indicators for an increased risk for HBOC.
- 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.
- CE programs are available for WHNPs to obtain evidence-based knowledge and competencies to provide HBOC counseling and genetic testing.

References

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Web resources

- A. nsgc.org/page/find-a-gc-search
- B. cancer.gov/cancertopics/genetics/directory

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