



Part I: Hereditary breast and ovarian cancer screening tool to identify patients who are at an increased risk of having a hereditary breast cancer syndrome

April 2025

Part I of this tool is used to screen for individuals who are at an increased risk of having a hereditary predisposition to breast cancer including but not limited to hereditary breast and ovarian cancer (HBOC) syndrome caused by pathogenic /likely pathogenic variants in the BRCA1 and BRCA2 genes.

Part II of this tool is used to identify individuals who are at increased risk to carry a pathogenic /likely pathogenic variant in BRCA1, BRCA2 or other hereditary cancer genes and would benefit from a genetic assessment.

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Did any of your first-degree relatives (parent, sibling, child) have breast or ovarian cancer?

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Did any of your relatives have bilateral breast cancer?

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Did any man in your family have breast cancer?

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Did any woman in your family have breast and ovarian cancer?

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Did any woman in your family have breast cancer before the age of 50 years?

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Do you have 2 or more relatives with breast and/or ovarian cancer?

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Do you have 2 or more relatives with breast and/or bowel cancer?

Management: With 1 or more positive responses, discuss referral to genetics.



When possible, the affected individual in the family at highest probability to carry a pathogenic variant* is offered testing first in order to (1) maximize the likelihood of identifying a pathogenic variant (2) decreasing the likelihood of an uninformative result.

It is important to note that any individual of Ashkenazi Jewish or French-Canadian ancestry may be offered genetic testing only for the variants more commonly found in these groups. A negative result in this situation only rules out those ancestry-specific variants.

This POC tool is based on the Family History Screening-7 (FHS-7) (Ashton-Prolla et al 2009), which was designed for use in primary care settings and demonstrated an overall sensitivity of 97.0% and a specificity of 53.0% for HBOC syndrome. Overall, using as cut point one positive answer, the sensitivity and specificity of the instrument were 87.6% and 56.4%, respectively for hereditary breast cancer syndrome.



Part II: Red Flags to identify patients at high risk of hereditary breast and ovarian cancer most likely to benefit from referral to genetics

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Eligibility criteria for genetic testing vary across Canadian provinces. In general, criteria are based on personal and/or family history that increase the likelihood of a hereditary cancer susceptibility syndrome.

These are criteria to identify individuals at high risk for hereditary breast and ovarian cancer (HBOC) syndrome. Strongly consider referral for a genetic assessment to your local genetics centre or hereditary cancer program if your patient has a family or personal history of:



Breast cancer at a young age (≤ 45 years)



Breast cancer ≤ 50 years and a limited family structure
e.g. adoption, few close relatives



Triple negative breast cancer diagnosed ≤ 60 years of age



Male breast cancer at any age



Ovarian cancer at any age



Multiple primaries in the same individual
e.g. breast and ovarian cancer, two primary breast cancer diagnoses



Breast or ovarian cancer at any age AND one or more (≥ 1) close relative(s) diagnosed with breast, ovarian, pancreatic or prostate cancer, on the same side of the family

*Closely related individuals typically refers to first- and second-degree blood relatives on the same side of the family



Ashkenazi Jewish ancestry AND one criterion below:



Personal history or close relative with breast, prostate, or colorectal cancer and/or GI polyposis at any age



Family history on the Ashkenazi Jewish side, that includes one or more (≥ 1) first- or second-degree relative(s) with breast, ovarian, pancreatic or metastatic prostate cancer, gastrointestinal polyposis at any age, or colorectal cancer diagnosed < 60 years



A blood relative with a known pathogenic/likely pathogenic genetic variant in a cancer susceptibility gene

Report or documentation containing specific gene and variant should be available.



An established risk model* calculating a 5 percent or greater ($\geq 5\%$) chance to carry a pathogenic/likely pathogenic variant in a gene associated with HBOC (e.g. BRCA1, BRCA2, CHEK2, PALB2, ATM) using family and personal history

Individuals who meet one or more of the criteria above may be eligible for high-risk breast cancer screening with MRI, genetic counselling and/or genetic testing.

*CanRisk Web Tool <https://www.canrisk.org/> - Free registration required;

*IBIS Breast Cancer Risk Evaluation Tool - Free download required <https://ems-trials.org/riskevaluator/>