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Genetics Education Canada: Knowledge Organization
Centre d'éducation en génétique canadien: Connaissances organisées
POINT OF CARE TOOL

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

July 2023

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 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 variant* in BRCA1, BRCA2 or other hereditary cancer genes and could therefore eligible for genetic counselling or genetic testing.

- ☐ Did any of your first-degree relatives (parent, sibling, child) have breast or ovarian cancer?
- ☐ Did any of your relatives have bilateral breast cancer?
- ☐ Did any man in your family have breast cancer?
- ☐ Did any woman in your family have breast and ovarian cancer?
- ☐ Did any woman in your family have breast cancer before the age of 50 years?
- ☐ Do you have 2 or more relatives with breast and/or ovarian cancer?
- ☐ 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.

***Pathogenic variant refers to a variation or change in a gene that has been established to be harmful to the gene's function. Formerly called a mutation.**

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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 clinical features 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 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 (≥ 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/