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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

Part I of this tool is used to screen for individuals who are at an increased risk of having a hereditary breast cancer syndrome including but not limited to hereditary breast and ovarian cancer (HBOC) syndrome caused by mutations in the *BRCA1* and *BRCA2* genes. Part II of this tool is used to identify individuals who are at high risk to carry a mutation in the *BRCA1* or *BRCA2* genes and are therefore eligible for genetic counselling or genetic testing.

1. Did any of your first degree relatives (parent, sibling, child) have breast *or* ovarian cancer?	Yes ❒	No ❒
2. Did any of your relatives have bilateral breast cancer?	Yes ❒	No ❒
3. Did any man in your family have breast cancer?	Yes ❒	No ❒
4. Did any woman in your family have breast *and* ovarian cancer?	Yes ❒	No ❒
5. Did any woman in your family have breast cancer before the age of 50 years?	Yes ❒	No ❒
6. Do you have 2 or more relatives with breast *and/or* ovarian cancer?	Yes ❒	No ❒
7. Do you have 2 or more relatives with breast *and/or* bowel cancer?	Yes ❒	No ❒

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

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 syndromes.

Reference: Ashton-Prolla P, Giacomazzi J, Schmidt AV, *et al*. Development and validation of a simple questionnaire for the identification of hereditary breast cancer in primary care. *BMC Cancer* 2009; 9:283

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Updated July 2019

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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

These are general guidelines to identify individuals at **high risk** for hereditary breast and ovarian cancer (HBOC) syndrome. You should consider referring your patient to your local genetics centre or hereditary cancer program for further assessment if they have a family or personal history of:

- Breast cancer diagnosis at a young age (<35-45 years) [both invasive and ductal carcinoma *in situ*]
- Ovarian cancer at any age [epithelial]
- Male breast cancer
- Multiple primaries in the same individual e.g. bilateral breast cancer (particularly if the diagnosis was before age 50), breast and ovarian cancer
- Breast cancer diagnosis **AND** a family history of two or more additional HBOC- related cancers, including breast, ovarian, prostate (Gleason ≥7) and pancreatic cancer
- High risk ethnicity (Ashkenazi Jewish, Icelandic) and a personal and/or family history of breast, ovarian or pancreatic cancer
- Triple negative breast cancer diagnosed <age 60

OR if s/he has a personal

- Probability of 10% or higher to carry a *BRCA* mutation

Eligibility criteria for genetic testing vary among organizations. In general, criteria are based on clinical features that increase the likelihood of a hereditary cancer susceptibility syndrome.

If possible, the affected individual in the family at highest risk to carry a mutation is offered testing first in order to maximize the likelihood of detecting a mutation.

Testing an unaffected individual should only be considered if an affected individual is not available for testing. There are significant limitations to interpretation of test results in an unaffected individual. Unaffected individuals can be referred for genetic counselling, risk assessment and information. It is important to note that any individual of Ashkenazi Jewish ethnicity or French Canadian ethnicities can be offered genetic testing for the mutations commonly found in these ethnic groups (e.g. three common mutations in those of Ashkenazi Jewish ethnicity). A negative result in this situation only rules out those ethnic-specific mutations.

For more information on Hereditary Breast and Ovarian Cancer such as screening recommendations and references see the complete *GEC-KO Messenger* at www.geneticseducation.ca

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