Part I: Hereditary breast and ovarian cancer referral screening tool to identify patients most likely to benefit from referral to genetics

Part I of this tool is used to predict which individuals should be referred for genetic counselling due to increased risk for a hereditary breast cancer syndrome including but not limited to hereditary breast and ovarian cancer (HBOC) syndrome caused by mutations in BRCA1 and BRCA2 genes. Part II of this tool is used to identify individuals who are at high risk to carry a mutation in BRCA1 or BRCA2 genes.

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.

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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 patients 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 s/he has 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