GENETIC TESTING: benefits and risks

Benefits
- Those who test positive can take steps to reduce risk.
- Test results for an individual can help their family members determine which mutations to test for.
- A negative test result may provide a sense of relief and prevent unnecessary screening, medications, or surgeries.

Risks
- Results may affect a person’s emotions, family relationships, finances, and privacy.
- While genetic information cannot lawfully be used to deny someone health coverage or employment, the law does not cover life, disability, or long-term care insurance, and it only applies to asymptomatic individuals (not those with a personal history of HBOC-related cancer).
- Treatments such as prophylactic surgery have risks and possible serious long-term complications.
- Ambiguous results, such as genetic variants of uncertain significance (VUS), may leave patients feeling unsettled or unsure about options.

Screening for hereditary breast and ovarian cancer (HBOC)

Many conversations about hereditary breast and ovarian cancer (HBOC) start with a primary care physician, who determines whether to refer someone to a genetic counselor. Alternatively, some people go straight to a genetic counselor. Regardless, a person’s risk for HBOC can be assessed through several steps as outlined below.

1: Family history risk assessment

The U.S. Preventive Services Task Force (USPSTF) recommends that primary care providers use a brief family history risk assessment tool to screen women with a personal or family history of breast, ovarian, tubal, or peritoneal cancer or who have an ancestry associated with BRCA gene mutations, such as Ashkenazi or Eastern European Jews. (See the separate fact sheet, “Understanding Risk Factors,” for more information on when to recommend screening.) Tools referenced by the USPSTF include:

- Ontario Family History Assessment Tool
- Manchester Scoring System
- Referral Screening Tool
- Pedigree Assessment Tool
- 7-Question Family History Screening Tool
- International Breast Cancer Intervention Study instrument (Tyrer-Cuzick)
- Brief versions of BRCAPRO

It is important to note that the screening results from different tools might not be consistent because the criteria and algorithms vary. Also, the tools may not identify some individuals with BRCA1 or BRCA2 mutations.

2: Genetic counseling

According to the USPSTF recommendation, individuals identified as high-risk through a familial risk assessment should be referred for genetic counseling. Genetic counselors are trained to:

- Take detailed family health histories.
- Assess hereditary patterns.
- Help individuals and families make decisions about genetic testing that are right for them, including which family member should be tested first.

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3: Genetic testing

If a person decides to have genetic testing, the genetic counselor can:

- Interpret and explain the results and provide emotional support.
- Explain medical management options (See separate fact sheet, “Preventive options for individuals diagnosed with HBOC.”)
- Provide referrals to medical specialists and support resources.
- Address concerns about the privacy of personal genetic information.

Providers should be aware that direct-to-consumer tests are not equivalent to genetic testing. In many cases, the results need to be confirmed in a Clinical Laboratory Improvement Amendments (CLIA)-certified laboratory before being used for clinical care.

Insurance coverage for genetic testing

The Affordable Care Act (ACA) requires non-grandfathered health plans to cover, without cost sharing, preventive services with a U.S. Preventive Services Task Force (USPSTF) rating of “B” or higher, which includes the BRCA testing recommendation. Importantly, genetic counseling, if appropriate, is covered without cost sharing by many health plans under the Affordable Care Act when used in accordance with the USPSTF recommendation. And the U.S. Department of Health and Human Services has advised insurance companies to cover the BRCA test without cost sharing when used in accordance with the USPSTF recommendation.

The Centers for Medicare and Medicaid Services (CMS) does not cover the USPSTF BRCA recommendation under the ACA. But multiple CMS Local Coverage Determinations (LCDs) on BRCA1 and BRCA2 Genetic Testing allow for regional coverage of BRCA genetic counseling and testing for individuals with personal histories of breast, ovarian, and other cancers that fit specific criteria for increased risk for a BRCA mutation. Some LCDs include coverage for individuals with certain strong family histories of breast, ovarian, and other cancers. However, CMS currently does not recognize genetic counselors as healthcare providers. This means that genetic counselors cannot be reimbursed for their services or see Medicare recipients without a physician or nurse practitioner present, unless the patient pays for services out-of-pocket.

Additional tools

For additional tools and resources, including those listed below, please visit: conversationsaboutcancer.org/provider/hboc

- Fact sheet: Understanding risk factors for hereditary breast and ovarian cancer (HBOC)
- Fact sheet: Screening for hereditary breast and ovarian cancer (HBOC)
- Fact sheet: Preventive options for individuals diagnosed with BRCA1 or BRCA2 mutations
- Other resources, including videos, conversation simulations, and more

2 - MCD Search Results (cms.gov)
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